ONE HUNDRED SECOND CONGRESS

Congress of the United States
House of Representatives

GOVERNMENT INFORMATION, JUSTICE, AND AGRICULTURE
SUBCOMMITTEE
OF THE
COMMITTEE ON GOVERNMENT OPERATIONS
B-349-C RAYBURN HOUSE OFFICE BUILDING
WASHINGTON, DC 20515

Public Hearing
Domestic and International Data Protection Issues:
Possible Uses and Misuses of Genetic Information

Thursday, October 17, 1991 at 9:30 a.m.
2154 Rayburn House Office Building

WITNESS LIST

Panel I

Dr. Bernadine Healy
Director
National Institutes of Health

accompanied by

Dr. James D. Watson
Director
National Center for Human Genome Research
National Institutes of Health

Dr. French Anderson
Chief, Molecular Hematology Branch
National Heart, Lung, Blood Institute
National Institutes of Health

Dr. David Galas
Associate Director
Office of Health and Environmental Research
Department of Energy

Panel II

Dr. Nancy Wexler
President
Heredity Diseases Foundation
New York, NY

Dr. Paul Billings
Division of Genetic Medicine
California Pacific Medical Center
San Francisco, CA

Mr. Jeremy Rifkin
President
Foundation on Economic Trends
Washington, DC

Dr. Philip Reilly
Executive Director
Shriver Center for Mental Retardation
Waltham, MA
On behalf of the American Society of Human Genetics

Opening Statement

CHAIRMAN BOB WISE

Subcommittee on Government Information, Justice, and Agriculture

Domestic and International Data Protection Issues: POSSIBLE USES AND MISUSES OF GENETIC INFORMATION

October 17, 1991

Today we will hold the second in a continuing series of hearings on domestic and international data protection issues. Data protection refers to the control of the collection, use, and dissemination of personal information. This is a more precise way of referring to privacy concerns.

The subject of the first hearing in April was public and corporate reactions to privacy. We considered the results of a Lou Harris poll of consumer attitudes on privacy issues. We also took a look at the reasons for the demise of Lotus Marketplace, a commercial product that would have made data on 120 million households widely available on CD-ROM.

At a hearing in the 101st Congress, we reviewed the direct marketing industry, and the use of bar code scanning technology to collect detailed, identifiable information on purchases made in supermarkets.

Earlier testimony made it clear that the right to privacy is routinely threatened by the increasing maintenance of detailed personal data by government and private sector record keepers. Consumers have little knowledge about how their personal information is collected and used, and they have little control over the way that the data is employed.

One theme that has emerged from these hearings is that new technology permits the maintenance and sharing of personal information that was never previously available. Those who market goods and services to consumers have a voracious appetite for personally identifiable data.

As a consequence, in the not too distant future, consumers face the prospect that a computer somewhere will compile a record about everything they purchase, every place they go, and everything they do. The information may be used to send targeted mail, to make telephone solicitations, to create consumer profiles, and for other purposes. If genetic information becomes available to marketers, the potential for invasion of privacy may be unlimited.

Another lesson we have learned is that it is important to establish rules for new types of personal information in advance. It is difficult to stop the use of data once it has started. It is much easier to prevent imprudent information practices before they begin.

Our focus today is on possible uses and misuses of genetic information. This is an especially complex and far-reaching area. The range of policy problems raised by the availability of identifiable genetic information goes far beyond traditional privacy concerns. We may also be required to reconsider the doctor-patient relationship, reexamine the basic principles of life and health insurance, and decide generally how we are going to treat individuals in light of the new information that will become available.

All of the traditional concerns about the use of personal information apply to genetic information. The standard privacy remedies of notice, access, and other fair information practices will be needed to protect the rights of individuals. But these traditional solutions will not be enough to address the broader problems that arise. We have to recognize in advance that the availability of identifiable genetic information may force us to find new ways of looking at privacy and at the relationship between individuals and society.

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One of the most serious and most immediate concerns is that genetic information may be used to create a new genetic underclass. People may be unable to obtain jobs and insurance, or participate in other routine activities, because of the stigma of having an undesirable gene. There is already evidence of discrimination on the basis of an individual's genetic makeup. We need to establish some clear rules about how this information may be collected, maintained, and used.

We may also need rules to keep genetic information from being used to fuel the consumer credit and direct marketing industries. I don't want to learn someday that people cannot qualify for credit cards because of their genetic history.

I don't want to learn that the mailing list industry is compiling lists of people who have genetic diseases or other genetic characteristics. Imagine the horrors of a direct mail campaign aimed at people with sickle cell trait, cystic fibrosis, or Huntington's disease.

As genetic testing becomes more sophisticated, the potential marketing possibilities will expand. If the genetic record demonstrates that a child has above average musical abilities, someone will try to sell the parents a piano. Another family might be told that genetic tests show their child is likely to be below average in math. So it is not too early to purchase a computer or enroll in a special school.

The possibility of such non-medical uses of genetic information are not entirely fanciful. There have been reports in the past that information on expectant parents has been sold to diaper services and others who sell products for the newborn. If genetic testing becomes widespread and if the data is not properly controlled, it may be commercialized in the same way. Someone needs to think about this before it is too late.

We are not going to solve all of these problems at today's hearing. The best we can hope is to begin the policy process by identifying the right questions and by considering how we should go about looking for solutions.

It is much to the credit of the Human Genome Project that the importance of these ethical, legal and social issues has been recognized and that funds are available for considering these issues. At the same time, I am concerned that the public policy problems are being approached in the wrong way and perhaps even by the wrong agencies. We may need to look for better ways to use the available funds. The manner in which we conduct scientific research may not be the best way to address public policy concerns. The problems are sufficiently complex that we may need an independent, comprehensive, and systematic review that will produce useful recommendations by a date certain.

There are other controversial aspects of the Human Genome Project that do not fall within the scope of today's hearing. For example, I am aware that there is a continuing controversy over the magnitude of the Human Genome Project. This is one of the so-called "big science" debates. It is not my intention to address this issue at this hearing other than to acknowledge its existence. I leave this aspect of the debate for other committees and other Members. For the purposes of this hearing, I accept the Human Genome Project as it is. Similarly, the many interesting and complex ethical issues raised by the use of gene therapy for medical treatment are issues for another day.

In closing, I want to acknowledge the pioneering effort on genetic privacy issues that has been made by John Conyers, Chairman of the Government Operations Committee, and ex officio member of this Subcommittee. Chairman Conyers introduced what I believe is the first bill on genetic privacy. H.R. 2045 proposes to establish rules governing use of genetic information by the federal government. This bill has been very valuable in raising the possibility that legislation will be needed to deal with genetic information issues.

REMARKS OF CHAIRMAN JOHN CONYERS, JR.

AT HEARING ON GENETIC INFORMATION, OCTOBER 17, 1991

MR. CHAIRMAN, I COMMEND YOU FOR SCHEDULING THIS HEARING ON "THE POSSIBLE USES AND MISUSES OF GENETIC INFORMATION" WHICH WAS DEVELOPED BY THE JOINT EFFORTS OF THE STAFF OF THE SUBCOMMITTEE AND THE FULL COMMITTEE.

THIS IS A CRITICAL ISSUE THAT MAY WELL BE THE NEXT MAJOR FOCUS OF THE FIGHT FOR INDIVIDUAL RIGHTS. LIKE DISCRIMINATION BASED ON RACE, GENETIC DISCRIMINATION IS WRONG BECAUSE IT IS BASED ON HEREDITARY CHARACTERISTICS WE ARE POWERLESS TO CHANGE.

THE FEAR IN THE MINDS OF MANY PEOPLE IS THAT GENETIC INFORMATION WILL BE USED TO IDENTIFY THOSE WITH "WEAK" OR "INFERIOR" GENES, WHO WILL THEN BE TREATED AS A "BIOLOGICAL UNDERCLASS."

THESE FEARS ARE BASED ON A LONG HISTORY IN THIS COUNTRY AND ELSEWHERE OF EFFORTS TO CREATE A "SUPERIOR" RACE OF PEOPLE AND TO TREAT OTHERS AS SECOND-CLASS CITIZENS:

-- IN AMERICA, THE EUGENICS MOVEMENT PERSUADED CONGRESS TO PASS THE IMMIGRATION ACT OF 1924, WHICH DENIED ENTRY INTO THE U.S OF CERTAIN PEOPLE DEEMED TO BE GENETICALLY INFERIOR.

-- AN EXTREME CASE OF MISUSE OF GENETIC INFORMATION WAS IN NAZI GERMANY, WHERE HITLER'S EFFORTS TO CREATE A RACE OF "PURE" ARYANS, RESULTED IN THE MURDER OF SIX MILLION JEWS.

-- IN THE LATE 1970'S, THE UNITED STATES AIR FORCE DENIED BLACKS WHO TESTED POSITIVE FOR SICKLE CELL ANEMIA THE OPPORTUNITY TO BECOME PILOTS -- THIS PRACTICE ENDED ONLY AFTER A FEDERAL LAWSUIT WAS BROUGHT.

-- A SURVEY BY THE OFFICE OF TECHNOLOGY ASSESSMENT FOUND THAT 20 "FORTUNE 500" COMPANIES HAD SOME FORM OF GENETIC SCREENING OF THEIR EMPLOYEES IN THE PAST 20 YEARS.

GIVEN THIS HISTORY, PUBLIC RELEASE OF PEOPLE'S GENETIC INFORMATION IS A "PANDORA'S BOX" THAT IS BEST LEFT UNOPENED.

IT IS POSSIBLE, THOUGH, THAT IN THE FUTURE GENETICS MAY BE SEEN AS A GREAT EQUALIZER -- "NOBODY IS PERFECT AND EVERYONE HAS DEFECTS" -- AND EVERYONE WILL UNDERSTAND THEY SUFFER FROM DISEASES AND BURDENS WHICH ARE DETERMINED BY THEIR GENES.

ONE OF OUR GREATEST CHALLENGES WILL BE TO EDUCATE PEOPLE ABOUT THE TRUE NATURE OF GENETIC INFORMATION, SO THAT IT DOES NOT BECOME THE BASIS FOR RACIAL HATRED AND DISCRIMINATION.

IN MY VIEW, THERE ARE THREE PRINCIPAL LEGAL QUESTIONS FOR THIS HEARING TO EXPLORE. 1) HOW TO PROTECT THE CONFIDENTIALITY OF GENETIC INFORMATION. 2) HOW TO PROTECT PEOPLE FROM DISCRIMINATION IN EMPLOYMENT. 3) HOW TO PROTECT PEOPLE FROM DISCRIMINATION IN ACCESS TO HEALTH, DISABILITY AND LIFE INSURANCE.

CONFIDENTIALITY

AS YOU KNOW, MR. CHAIRMAN, I HAVE INTRODUCED LEGISLATION, THE HUMAN GENOME PRIVACY ACT (H.R. 2045) , WHICH PREVENTS DISCLOSURE OF GENETIC RECORDS WITHOUT AN INDIVIDUAL'S PERSONAL WRITTEN CONSENT, AND GUARANTEES EVERYONE THE ABILITY TO CORRECT OR AMEND HIS OR HER RECORDS THAT ARE MAINTAINED BY THE FEDERAL GOVERNMENT AND ITS GRANTEES AND CONTRACTORS.

THIS IS A FIRST STEP IN A LEGISLATIVE MOSAIC WHICH WILL ULTIMATELY PROVIDE THE BROADEST POSSIBLE PROTECTION AGAINST ABUSE OF THIS INFORMATION WHICH IS THE MOST SENSITIVE INFORMATION ABOUT A PERSON.

MY BILL ATTEMPTS TO SET FORTH PRINCIPLES THAT SHOULD GOVERN THIS AREA THAT CAN THEN BE USED TO EXTEND PRIVACY PROTECTION TO THE PRIVATE SECTOR.

EMPLOYMENT DISCRIMINATION

I AM VERY SERIOUSLY CONCERNED BY THE POTENTIAL FOR ABUSE IN THIS AREA. JUST LAST WEEK, FOR INSTANCE, THE OFFICE OF TECHNOLOGY ASSESSMENT RELEASED A STUDY SHOWING THAT 59% OF EMPLOYERS REQUIRE PRE-EMPLOYMENT HEALTH EXAMINATIONS OF ALL OR MOST JOB APPLICANTS.

WILL THESE TESTS BE USED BY COMPANIES TO REJECT PEOPLE WITH A PREDISPOSITION TO CORONARY DISEASE OR TO ALCOHOLISM OR TO DEPRESSION OR CANCER, ON THE GROUNDS THAT GENETIC SCREENING IS HELPFUL TO HIRING A "PROBLEM-FREE" WORKFORCE THAT WILL MAXIMIZE PRODUCTIVITY?

I AM DISAPPOINTED THAT THE REGULATIONS RECENTLY PUBLISHED BY THE EQUAL EMPLOYMENT OPPORTUNITY COMMISSION DO NOT EXPLICITLY STATE THAT DISCRIMINATION ON THE BASIS OF GENETIC INFORMATION IS A VIOLATION OF THE AMERICANS WITH DISABILITIES ACT.

I WILL BE SUBMITTING SOME SPECIFIC QUESTIONS TO EEOC CHAIRMAN EVAN KEMP TO FIND OUT WHY GENETIC DISCRIMINATION WAS NOT PROTECTED IN THE REGULATIONS. IF NEW LEGISLATION IS NEEDED, WE MAY HAVE TO BEGIN CONSULTATIONS AND FILL ANY GAPS IN THE LAW.

INSURANCE DISCRIMINATION

WE WILL HEAR TESTIMONY TODAY THAT MANY PEOPLE HAVE BEEN DENIED COVERAGE OR CHARGED HIGHER PREMIUMS AS A RESULT OF GENETIC INFORMATION IN THEIR MEDICAL RECORDS. THIS IS AN ALARMING PROBLEM THAT MUST BE ADDRESSED.

ONE SOLUTION IS TO PROHIBIT THE USE OF GENETIC INFORMATION TO DETERMINE ELIGIBILITY FOR HEALTH INSURANCE. A BILL TO THIS EFFECT HAS BEEN PASSED BY THE CALIFORNIA LEGISLATURE AND SENT TO THE GOVERNOR TO SIGN OR VETO IT IN THE NEXT 30 DAYS.

I AM ALSO VERY INTERESTED IN HEARING FROM THE WITNESSES WHETHER THIS PROBLEM CAN ALSO BE RESOLVED BY ENACTING A NATIONAL HEALTH INSURANCE PLAN WITH UNIVERSAL COVERAGE. IF WE AS A SOCIETY ACCEPT THE RESPONSIBILITY FOR PROVIDING ADEQUATE HEALTH COVERAGE AS A FORM OF SOCIAL INSURANCE, THOSE WITH A HIGHER INCIDENCE OF ILLNESS OR A HIGHER RISK FOR ILLNESS WOULD NOT HAVE TO BE VICTIMS OF STEEP INSURANCE PREMIUMS OF POSSIBLE EXCLUSION FROM THE WORKPLACE.

CONCLUSION

MANY OF US IN CONGRESS WERE EXTREMELY CONCERNED ABOUT THE POSSIBILITY OF ABUSE OF GENETIC INFORMATION WHEN CONGRESS DECIDED TO SPEND MORE THAN $3 BILLION OVER THE NEXT 15 YEARS TO FUND THE HUMAN GENOME MAPPING PROJECT.

ONE RESULT OF THAT CONCERN WAS THAT 3% OF THE FUNDS FOR THE HUMAN GENOME MAPPING PROJECT WERE TO BE ALLOCATED TO STUDY THE "ETHICAL, LEGAL AND SOCIAL IMPLICATIONS" OF GENETIC RESEARCH. WE WILL HEAR TESTIMONY TODAY ON WHETHER THAT MONEY IS BEING SPENT IN A USEFUL MANNER.

THE USE OF GENETIC INFORMATION RAISES FEARS IN THE MINDS OF MANY PEOPLE THAT THIS INFORMATION WILL BE USED AGAINST THEM. THE CHALLENGE THAT WE FACE IN CONGRESS IS TO MAKE SURE THE LAWS ARE ADEQUATE TO PROTECT ALL OF US. I LOOK FORWARD TO THE TESTIMONY TODAY THAT WILL HELP US REACH SOUND JUDGEMENTS ON SOME OF THESE VERY DIFFICULT ISSUES. THANK YOU, MR. CHAIRMAN.

For Release on Delivery

Testimony by

Dr. Bernadine Healy, Director
National Institutes of Health
Public Health Service
Department of Health and Human Services

Hearing on the Possible Uses and Misuses of Genetic Information

Before the

Subcommittee on Government Information, Justice, and Agriculture
Committee on Government Operations
House of Representatives

October 17, 1991

Mr. Chairman and members of the subcommittee, as the federal government committed itself to invest heavily in science over 50 years ago, it did so in the context of the Vannevar Bush tenet that science cannot live by and unto itself alone. The mission of the National Institutes of Health (NIH) is to bring science to the benefit of humankind by discovering and uncovering fundamental new knowledge of living organisms and translating them into human benefits. In this, we have a proud history of success. Indeed, Americans and their families are intimately touched by the triumphs of biomedical science in very deep and personal ways almost on a daily basis. That very success combined with the evident power of the knowledge of living organisms obligates us to address the social implications of our triumphs as well.

I believe future historians will point to the last decades of the 20th Century as the time when America helped the life sciences come of age. Looking at biomedicine in the 1990's, historians should see both new strengths and skills and a mature commitment to use science responsibly in helping others.

Under the auspices of public institutions like NIH, the growth of knowledge about biology has been inspired by its applications to human health. A newly discovered molecule is given biological meaning by the bench scientist, and it is put to good use at the bedside by the clinical scientist. Two witnesses here today embody the translation of scientific discovery into new treatments probably better than any other I know. Dr. James Watson, who is director of the National Center for Human Genome Research (NCHGR), discovered in 1953, the structure of DNA, the hereditary molecule. He was later awarded a Nobel Prize for that work. In 1953, no one knew that genetic

information was inscribed in DNA or how genetic information was passed from one dividing cell to its progeny or even from parents to their children. Understanding the structure and function of specific genes seemed light years away. Even more distant was the idea that we would ever be able to do anything about the thousands of human diseases caused by faulty genes. But less than 40 years after Dr. Watson's discovery, Dr. W. French Anderson, of the National Heart, Lung, and Blood Institute (NHLBI), and his colleagues at the National Cancer Institute succeeded in transferring healthy genes into a patient suffering from one of those genetic diseases and seeing the genes work to help a severely ill child.

New knowledge about the nature of genes, how they act together, by themselves, and in concert with our environment, will no doubt influence the way we think and feel about ourselves and our fellow citizens. In my nomination statement to the Senate subcommittee, I spoke about the interface between biomedical science and the public interest. NIH must set its research priorities in response to public need, our very reason for being here. I said then that, "whatever we do in science is ultimately done in the context of society; whatever we do in biomedical research must be in the interest of the public....This is the history of recombinant DNA research and the oversight of the NIH Recombinant DNA Advisory Committee. This is the history of the Institutional Review Boards for overseeing any medical research that involves human participants. The same principle underlies the plans of the Human Genome Project to invest part of its resources into studies of the ethical implications of knowing a person's genetic make up. As we move ahead, these approaches are already serving as models for assuring the public that science indeed

does not live by and unto itself alone, but in the service of man- and womankind."

My goal today is to report to you how, in one important way, NIH in the 1990's intends to carry on its responsibility to look beyond the laboratory to the human consequences of our work. I would simply like to make five points about these issues, to describe to you NIH accomplishments in this area, as well as the steps we are taking to prepare for the future.

The Power of Medical Technologies to Do Good

First, we are driven by a deep commitment to the power of medical technologies to do good. One of the first technologies ever discovered and developed by humans is fire. By harnessing this force, people have been able to cook their food, regulate their living environment, melt ore to make metals, and develop transportation machines that can take you around the block or around the universe. Indeed, we cannot live our lives without it. All of us learned as children, however, to treat this force with respect, for we have all seen its awesome power to do harm.

The tools and discoveries of biomedical research deserve similar respect. Our abilities to harness the biological forces of nature have become extraordinary. Today, we have techniques that allow us to look directly at our genes and their precise chemical structures. DNA, the chemical that genes are made of, contains the information necessary for the development of a human being and to direct its physiological functions. In the future, efforts to understand human disease will focus increasingly on the role of genes.

With new research tools being developed by the Human Genome Project, scientists will be isolating genes at rates never before possible. It is estimated that human beings have some 100,000 genes in their DNA, but only a few thousand have been identified and even fewer have been isolated. As each gene is discovered, new insights unfold, and with them arise new prospects for diagnosis and treatment of previously baffling and untreatable conditions. For example, upon analyzing the cystic fibrosis (CF) gene and comparing it to other known genes, Dr. Francis Collins, at the University of Michigan, could immediately see that the CF gene was likely to have something to do with transporting substances across the cell membrane. As a result, Dr. Collins is working out ways to deliver genes to lung cells to enable them to produce the protein that is altered or missing in cystic fibrosis. The more genes we discover, the easier it will be to determine their functions in the human body.

An extraordinary example of how genetic knowledge has been translated into treatment for disease is the gene therapy story happening at the NIH this very moment. As part of an historic experimental procedure, two children, who were born with adenosine deaminase (ADA) deficiency, are being treated with their own blood cells, which have been genetically engineered to correct the disease. ADA is caused by altered genes that prevent their bodies from making a vital immune system protein. Without this protein, the children's immune systems were unable to protect them from infections. Dr. W. French Anderson and his colleague, Dr. Michael Blaese at the National Cancer Institute (NCI), devised a way to insert a functional ADA gene into the blood cells of these children. With this treatment, the children's immune systems are starting to function as they should.

Moreover, the blossoming of our knowledge about the molecular basis of human genetics has produced breakthroughs not just for rare genetic diseases, but for problems as common among our families as cancer and heart disease. I am also proud to report that methods designed by NCI's Dr. Steven Rosenberg to use gene transfer techniques to treat cancers have been approved to begin at NIH. Using this powerful technology, we have embarked on the first step of a long journey toward a human cancer therapy. Scientists have already begun to think of other types of diseases that may be treated by gene therapy as well. With the support of the National Institute of Diabetes, Digestive, and Kidney Diseases and the National Center for Research Resources, scientists are developing methods to deliver healthy genes to liver cells in hopes of treating familial high cholesterol, a condition that leads to early heart attack. Dr. Ronald Crystal, at NHLBI, is also experimenting with ways to deliver genes to lungs to treat a hereditary form of emphysema, which is caused by lack of the protein alpha-1-antitrypsin. None of these procedures is ready for use in humans, but they demonstrate the breadth of creativity that is inspired by gene discoveries.

Even before treatments are developed from a gene discovery, the knowledge is of enormous human value. When a disease gene is isolated, it enables us to better understand the basic cause of the disease as well as to develop tests to detect the altered gene in patients. Such tests are immediately useful for accurate diagnosis, especially when the biochemical basis of the disease is not understood. Although a diagnosis is not a cure, fundamental knowledge of cause is an essential step towards conceiving therapy and even cure. Moreover, even genetic tests indicating a risk for a disorder

can lead to changes in the person's nutrition or environment that may reduce that risk. Like fire, however, all powerful tools can be dangerous if misused or abused, and biomedicine's new molecular tools are no exception. As the biomedical sciences mature, biomedicine's social responsibilities also grow. Over the past 20 years, professional and public consideration of the ethical, legal, and social implications of science have become an integral part of the biomedical research process. The NIH has a long history of concern for the proper conduct of research on human participants and protection of the confidentiality of the information generated through biomedical research.

The NIH Commitment to Social Policy Implications of Science

A second matter that is derived from the first is that NIH, as the leader of biomedical research in the world, has a firm commitment to the social policy implications of science. NIH has the will and the ability to look ahead, reflect upon, and prepare for the social implications of biomedical research. Over the past two decades, the agency has established a strong record of its commitment to this responsibility. First, consider our human subjects guidelines. Beginning in the 1960's, NIH initiated the development of the rules that now protect the rights and interests of biomedical research volunteers. Clinical research carried out by investigators at the NIH and at institutions that receive NIH funds are now regulated by the Department of Health and Human Services' **Regulations for the Protection of Human Subjects (Title 45 CFR Part 46)**, which address the confidentiality of research information. The Department's **Office for**

Protection from Research Risks (OPRR), which is housed at the NIH, is charged with overseeing compliance of both intramural and extramural clinical research with the HHS regulations. It is led by Dr. Charles McCarthy, a man who has devoted a 20-year career at NIH to protecting the interests of human and animal research subjects. The policies he has helped to develop serve as a global model for human subjects research, and this year have been officially adopted by all branches of the federal government under the **Common Rule of 1991.**

Included within the general policies for protecting human research volunteers are rules that govern the confidential management of biomedical and behavioral research data, including genetic information as they relate to individual research subjects. The intent of these regulations is to ensure that research information that is publicly available cannot be linked to a specific individual by way of name, description, or other identifying element, without the consent of the research participant.

The human subjects policies call for local **Institutional Review Boards** at each investigator's clinical research facility. The IRB reviews research proposals to ensure they adequately protect human subjects, including their privacy. All applications submitted to the NIH are scrutinized for compliance with the human subjects regulations by the peer review process and by NIH staff. An additional protection, authorized by the **Public Health Service Act**, enables NIH to issue **Certificates of Confidentiality**, to investigators conducting federally funded clinical research that collects sensitive information about individuals. These Certificates protect research records from subpoena by courts or other enforcement agencies and from other types of compulsory disclosure. Certificates of Confidentiality are

particularly important for research that generates sensitive information about behavior disorders, such as manic-depression or substance abuse.

The **Privacy Act of 1974**, which pertains to federally controlled systems of records, provides confidentiality protection for biomedical research records about individuals in government facilities.

A particularly active element in the protection of research subjects within the NIH intramural research program is the **Bioethics Program** of the NIH Warren Grant Magnuson Clinical Center. This office, now led by Dr. Michelle Carter, an ethicist who brings to her job both a doctoral degree in moral philosophy and the practical experiences of a clinical nurse, provides consult services to NIH staff both in review of clinical protocols and in clinical decision making about the care of patients.

A second element of NIH's leadership in dealing with the social implications of science is the Recombinant DNA Advisory Committee (RAC). In the mid-seventies, NIH helped the scientific community proceed cautiously into the exciting field of DNA splicing. Under the leadership of Dr. Donald Frederickson, NIH established research guidelines and a review system which subsequently both public and private sector scientists have adopted. The **Office of Recombinant DNA Research** and the **Recombinant DNA Advisory Committee** have been instrumental in the responsible growth of America's leadership in biotechnology, and its principles and procedures have been replicated throughout the world.

In the early 1980's, for example, NIH used this process to conduct the most thorough and public assessment of a new research proposal ever performed in the history of biomedical science: the

review of the first human trials of gene therapy, conducted by Dr. Anderson and his colleagues, for inherited immune disease and now enjoying success at the NIH clinical center. I might also note that Dr. Anderson provides not only scientific and medical expertise to the field of gene therapy, he is now the editor of a new scientific journal, **Human Gene Therapy**, which in its brief existence has already published more articles examining the ethical and social policy dimensions of genetic medicine than the rest of the medical literature combined.

There are other examples of NIH's sensitivity to the social implications of science. In the late 1980s, NIH took the initiative to help society explore controversial science policy issues in biomedical research and the proper care of animals in research settings, and, through the National Center for Nursing Research, began its first funding program for extramural studies of ethical issues in the clinical delivery of health care.

Most recently, with the creation of the National Center for Human Genome Research, NIH established a precedent-setting research program to anticipate and address the ethical, legal and social implications of advances in human genetics, including implications for personal privacy. This program represents an important innovation in federal science planning: for the first time, federal support for a major new scientific initiative like the Human Genome Project is being complemented by concurrent work aimed at addressing the social implications of that science.

NCHGR's Ethical, Legal and Social Implications (ELSI) program is directed by Dr. Eric Juengst, a philosopher trained in ethics who comes to us from a career of working on genetics issues within the

biomedical community. He is aided by an active group of consultants in health policy, ethics, law, genetics, and psychology, led by Dr. Nancy Wexler, who is well known for her work in identifying the location of the Huntington's disease gene. In its first two years, the NCHGR Ethical, Legal and Social Implications (ELSI) program has provided over $6 million in support of 18 research projects, 8 education programs, and 10 public conferences. These include projects aimed at developing professional guidelines for the confidentiality of genetic records, clarifying the legal foundations of genetic privacy, assessing the social implications of genetic identification techniques, surveying collections of genetic data, and evaluating the potential uses of genetic information by insurers and employers. With NIH support, these studies are being channeled into the health policy-making process through deliberative and education efforts by organizations as diverse as the National Academy of Sciences, the Council of State Governments and the Alliance of Genetic Support Groups. In fiscal year 1992, NCHGR plans to spend 5 percent of its grant-making budget on such studies, or approximately $5 million, toward the program's five-year goal of developing sound policy options on this range of issues by 1995. We are proud to say this is the largest and most focused public investment in this form of biomedical ethical, legal and social impact assessment of science to date, and has already served as a model for similar activities by other national genome research programs around the world.

Two themes that are emerging from the Genome Center's work are particularly relevant to today's proceedings: the importance of individual control over the acquisition of personal genetic information and the need to improve social protection against genetic

discrimination. I would like to discuss these two matters in greater detail.

Why the Acquisition of Personal Genetic Information Must be Voluntary

First, individuals must have control over the acquisition of personal genetic information. Genetic information is especially personal because it can identify health risks we inherit from (and often share with) our families, and that are built into our bodies at a very basic biological level. At the same time, contrary to popular belief, the predictions that genetic analyses provide do not have crystal-ball qualities: they always involve uncertainties to weigh, and cannot predict how individuals will experience the possibilities they face. It is the right of individuals and their families to determine the relative value of this information for their own lives, and to control its acquisition as well as its disclosure to others.

Much of the energy of the genome Center's ethics program has been devoted to addressing the challenges of applying this principle in the clinical setting. As a matter of definition, genetic information almost always has implications for close relatives who may share genes. Consider the case of a pair of twins at risk of inheriting the gene that causes Huntington's disease. If one twin wants to know his status and the other doesn't, should the test be provided to the first? What are the clinician's duties to warn others who may be at genetic risk, or to protect those who would rather not know? As long as clinicians depend upon family linkage tests, which require testing

of multiple members of a family to obtain information about any single member, they will continue to face privacy dilemmas of this sort.

To develop guidance on these questions, the ELSI program, together with its counterpart at the Department of Energy, is supporting a two-year study of professional policy issues in genetic services by the National Academy of Science's Institute of Medicine. For example, in the case of the twins, some testing programs would go so far as to deny testing to the curious twin in order to protect his brother's right not to know. Other professionals would be satisfied to ensure simply that the reluctant brother did not learn his twin's status (and thus his own) from them.

Similar questions arise about the use of predictive genetic tests with children. This is one of the questions the National Cancer Institute and the National Center For Human Genome Research are currently considering in developing guidelines for the clinical uses of genetic tests for cancer risks. The tradition in medical genetics has been to reserve genetic testing for adults when the information to be obtained is only useful in health planning relevant to adult life. When tests are performed on children, it has been because the information can be of immediate use in preventing harm, as in the case of newborn PKU screening. Now, however, tests are being developed that fall into the middle ground: for example, tests that use genetic markers as measures of the risks people face of developing cancers. While these markers are not sure predictors of cancer, and there are no guaranteed preventive measures to take in response to these warning signals, there are sometimes dietary and other steps that can reduce the risks they signal. Since these steps are usually more effective

the earlier in life they are taken, should this form of genetic testing be recommended to families with a history of cancer?

Finally, it is worth noting the protection of genetic information not generated under Federal funding is much less clear than the protection that federally funded research records enjoy. Much genetic research depends on collections of information about families and the patterns in which they inherit certain traits. Such family information is held in a number of different ways: family medical records; state public health departments; genealogical organizations; and, increasingly, genetic disease support groups and voluntary health organizations. Since many of these collections currently enjoy no systematic set of privacy protection, the NCHGR ELSI program is creating a Privacy Task Force to develop model policies for collections of genetic data held by private-sector organizations, such as voluntary health associations or commercial DNA banks. This group will meet quarterly over the next year, to survey existing collections and identify and address common confidentiality problems through a set of guidelines for genetic information access and disclosure.

We Must Prohibit Discrimination on the Basis of Genotype

A second major ethical issue is to prohibit discrimination based on genotype. As scholars point out, "Genetics is the science of differences." Because genetic information has the potential to be used to categorize people unfairly, genetic information about individuals can also be stigmatizing, and has in the past exposed individuals to discrimination and the loss of access to social and economic opportunities. For example, during the introduction of

state-based screening programs for sickle cell disease during the 1970's, people were denied health insurance and jobs simply because they were identified as carrying one copy of the gene--a trait that has no significant implications for their health. We must never again allow that sort of confusion and discrimination to follow from the use of genetic services. This is particularly important in light of the current efforts to unravel the molecular genetics of neurological and behavioral disorders such as Alzheimer's disease, schizophrenia or alcoholism: conditions that already bear the burden of stigmatization in our society. Indeed, much of the interest in improving protection on the privacy of genetic information are aimed ultimately at preventing its abuse in this way. Discrimination on the basis of genotype alone is ethically no different from discrimination based on other biological traits such as gender, skin color, or disability, and should be prohibited by similar forms of civil rights protection.

Besides providing support for research and education on these issues, the genome Center's ethics program has taken the initiative, through the joint NIH-DOE ELSI Working Group to directly address several of the highest priority issues in this area. For example, to help prevent unwarranted genetic testing in the work place, the ELSI working group has provided comments to the Equal Employment Opportunity Commission, which regulates the newly enacted Americans with Disabilities Act. These suggestions seek to prevent job candidates from being excluded from jobs on the basis of mandatory, pre-employment genetic test results. They closely parallel the guidelines for employer testing put forth by the American Medical Association just two weeks ago.

In addition, the ELSI Working group has established an Insurance Task Force, a group representing the insurance industry, consumer groups, geneticists, health policy analysts, and insurance policy makers, which has set for itself the task of establishing industry guidelines and public policy options regarding insurer use of genetic information. As more people become aware of their risk for genetic disease, commercial insurers must decide whether to include such information in their underwriting decisions to address increased economic pressure by knowledgeable consumers and competitors who will use the information. Together with a NIH funded research team at the University of Florida, they are gathering information on current and potential insurance industry underwriting practices and plan to publish a report during the next year that will form the basis for developing collectively endorsed guidelines. The Insurance Task Force plans to have formal recommendations by the Fall of 1993.

Keeping Genetic Information in Context

Privacy issues that surround genetic information are akin to those surrounding other kinds of personal, medical information. In developing approaches to resolving privacy issues, it is important to place genetic analysis and information in its proper context within the spectrum of current biomedical capabilities. Privacy issues related to the acquisition and use of genetic test results are important because they epitomize the challenges of managing general medical information. In a growing number of areas, however, the boundaries that delineate genetic information from other kinds of medical information are fuzzy at best. As a physician, for example, I

can tell from a blood test whether a person has an inherited form of high cholesterol. A biochemical test will measure cholesterol, but the amount of cholesterol can tell me whether a patient has an inherited alteration in their LDL receptor gene. The commonly used test to detect the genetic metabolic disorder, phenylketonuria, is also a biochemical test that can tell us about a person's genetic make up. Should direct information about a person's DNA be regarded and treated any differently than these proxy tests for gene function?

We must not forget that many kinds of personal and medical information carry powerful stigma. Consider HIV status or the results of intelligence tests, for example. The notion of privacy itself has historically been defined by social and cultural attitudes about what kinds of information should be regarded as personal or secret. These attitudes often arise out of fear, justified or not, that knowledge of the information by others would result in personal, social, or economic harm. In considering laws and policies about the regulation of genetic information, we must be mindful not to add unwittingly to the historical taint such information has carried.

It is important too for the public to understand how genetic information fits into the greater social and medical context. In 1986, the Congressional Office of Technology Assessment commissioned a nationwide survey to assess public knowledge and attitudes about biotechnology, the broad field of science that includes DNA research and genetic testing. The survey found that while the public expressed concern about genetic engineering in the abstract, it approved of nearly every specific therapeutic application. Eighty-nine percent of the 1,273 people surveyed said they approve of making genetic tests available through doctors. Eighty percent say they expect

developments in science and technology to benefit them and their families. And finally, when faced with a choice between the risks and benefits to society, 62 percent feels that the benefits outweigh the risks.

NIH is currently developing a strategic plan designed to chart the agency's course into the next century. A prominent theme in that planning has been the recognition that ethical, legal and social challenges of the sort now being addressed for genetics can also attend new scientific advances across the biomedical disciplines. In keeping with the agency's tradition of attempting to address these kinds of science policy issues proactively, the NIH is establishing a new center for science policy studies. By anticipating emerging issues and providing a hub for the activities of NIH institutes and centers, and the discussions of the scientific community, the Center will stand, for future historians, as an emblem of the ethical integrity of biomedicine at the dawn of its adult career.

Summary

In summary, I will reiterate the five points I would like to leave with you today:

First, the biological revolution has extraordinary power to do good. As long as the use of our new genetic knowledge is guided by the traditional ideals of the healing professions--to help improve the human condition without doing harm--we can expect real benefits to come of it.

Second, however, we need to prepare for the fact that, like all powerful tools, genetic information can be misused and abused. As I

hope to have illustrated, NIH has the will and ability to work with the American public and the scientific community in preparing for the responsible use of genetic information for now and the future. To date, the best example of that will is the precedent-setting work being conducted in concert with the human genome program through our ELSI program.

Third, as a part of that work, it will be imperative to continue to protect the voluntary nature of genetic services. The rights of people to determine for themselves whether or not to pursue genetic information about themselves must be defended, even from the forced choices that discriminatory social practices may create.

Fourth, in order to allow those who choose to do so to benefit from our new genetic tools, discrimination based on genotype must be prohibited as a matter of basic civil rights.

And finally, in the bright light of the Human Genome Project and its ELSI program, it is important not to let genetics eclipse the important ethical, legal and social issues that attend other biomedical advances. It is the purpose of NIH's new center for science policy studies to illuminate this broader view of the road ahead.

Thank you. I will be happy to answer any questions you might have.

Statement of

David J. Galas
Associate Director for
Health and Environmental Research
Office of Energy Research
Department of Energy

before the

United States House of Representatives
Committee on Government Operations
Subcommittee on Government Information, Justice and Agriculture

October 17, 1991

Statement of

David J. Bates

Associate Director for

Health and Environmental Research

Office of Energy Research

Department of Energy

before the

United States House of Representatives

Committee on Government Operations

Legislation on Government Information, Justice and [illegible]

October 17, 1991

Mr. Chairman and Members of the Subcommittee:

I am pleased to be here today to discuss an extremely important issue. This issue concerns how we deal with both the benefits and the potential for misuse inherent in the enormous increase in human genetic information that will result from the Human Genome Program. What are the ethical, legal and social implications of this new information about ourselves and our individual genetic constitution, and what should we be doing to prepare for its use? My testimony will focus on the nature of some of these issues, and the role of the Department of Energy (DOE) in examining some of these issues as part of the Human Genome Program. This country's Human Genome Program is carried out by DOE together with the National Institutes of Health (NIH).

It is clear that the Human Genome Program will provide new tools for the practice of medicine and, in doing so, will provide significant opportunities to improve the health of people worldwide. Because of the genetic information produced by the Human Genome Program, both the diagnosis and treatment of human disease will improve radically and will markedly change the practice of clinical medicine in the 21st century. In the future, the ability to detect genes that can predispose an individual to a certain disease will shift the basic philosophy of medicine from a reactive philosophy, aimed at curing sick patients, to a preventive philosophy focused on keeping people well. With that fundamental shift, many more people will be able to live normal, healthy lives.

Of the roughly 100,000 human genes, many have been identified and mapped, but the functions and map locations of 97 percent of them are still to be discovered. The pace of identification and characterization of human genes, however, is increasing sharply because of the Human Genome Program. In future medical practice, thousands of an individual patient's genes, many yet to be discovered, could be characterized and an accurate profile of the subject's medical state will thereby be drawn to allow patient and doctor to act to prevent potential problems. Once a gene is implicated as the cause of a disorder, the door is opened to finding a cure for the disorder, or at least for the mitigation of medical problems. After the discovery of such a gene, the knowledge gained will in most cases lead to an understanding of the biochemical nature of the problem and, subsequently, to potential medical solutions.

In the next century it may be possible to analyze 50 or more genes from the DNA of an infant to detect the various forms that can predispose the child to many common diseases (cardiovascular, cancer, autoimmune, metabolic, etc.). The genetic basis for particular predispositions to heart disease and some cancers is already known. The catalog of so-called genetic diseases is long and growing. What is becoming clear is that most diseases have a genetic component - what we now call a genetic disease simply means that the genetic component is large enough for us to discern with our relatively crude methods. With newly forged tools, the genetic component of more and more common disorders will be revealed.

The intimate genetic knowledge that will ultimately be available as a result

of the Human Genome Program is comparable to a patient's medical records today. Although much more detailed and predictive, genetic records will be subject to potential abuses, just like today's medical records. Almost all the ethical, legal and social issues raised by the future prospects of genetics apply to current medical practice, but many of these long-standing issues have never been properly addressed and will be of increasing concern as genetic knowledge grows. Indeed, some of the ethical and legal issues of genetic knowledge are already with us today. The genome program is providing us with the opportunity and motive to address in a systematic fashion some of the long-standing policy problems that are associated with the use of genetic information as well as anticipate new issues that might arise.

This information, with its abundant beneficial uses, could also be used in ways that could result in discrimination against individuals, invasion of individual privacy or misallocation of health-care resources. DOE and NIH consider it essential to anticipate and avoid those possible consequences of the new genetic knowledge, rather than simply to react when they occur. Therefore, we have instituted a research and education program, as an integral part of the Human Genome Program, dealing with the ethical, legal and social implications of the increased knowledge of the human genetic make-up.

Before discussing this activity in some detail, I would like to briefly describe here some of the relevant work being done in the Human Genome Program to indicate the scientific underpinnings of the issues.

Genetic Information from the Genome Program

The Human Genome Program's single principal goal is the complete mapping and sequencing of the entire complement of genetic information in the human body -- the human genome. Mapping means determining the positions of genes on the chromosomes, and sequencing means determining the precise chemical structure of the DNA. This complete set of genetic instructions, called the genome, controls the development of a complete organism from a single cell, the constant maintenance and repair of its parts, and the growth and metabolism of an organism. Understanding this intricate and largely unknown genetic blueprint, written in the genetic language of DNA and encoded in the 24 different human chromosomes, has now become the center of the Human Genome Program.

This program is a focused research effort that departs from the norm for basic biological research in several ways. It is the largest coordinated effort in biology ever directed at a single goal, and the first major interdisciplinary project in biology. It is also the first project in biology that has technology development, for mapping and sequencing and for automation of laboratory analysis, as an integral part of the program. Technology development is, in fact, a particular emphasis of the Department of Energy's efforts in the program. The program in all its complexity has been well described elsewhere. I will confine my remarks here to describing some of the research that directly affects the public.

Once a gene has been located and characterized, a simple test can be devised to detect each of its different forms, one or more of which may have a

detrimental effect. If one of those forms has a medical implication for the person who carries it, then this test itself has immediate medical consequences. Many of the known "genetic" diseases are determined by a single gene (cystic fibrosis, Tay-Sachs, sickle-cell anemia, for example), but most genetically related medical problems are determined by the states of many different genes - these are generally called multi-genic disorders. For these latter disorders a medical conclusion would require tests for many gene variants.

Because of the new DNA technology, information on the state of any particular gene can be obtained from an individual patient from a simple blood test. Information of this kind can be compiled and kept in a patient's records. The rules of genetic inheritance enable one to calculate the probabilities of particular states of particular genes being carried by members of the patient's family. This is the one way in which genetic information is different from other sorts of medical information. Information can be obtained about other family members from information about the tested individual.

There are additional genetic facts to keep in mind when considering societal issues discussed later in this testimony. Not only do some genetic effects cause increased susceptibility to disease rather than the disease itself, but some disease states are only manifested later in life - the so-called late-onset disorders. The genetic knowledge in some of those cases predicts that a person who is now completely normal and fully functional will be stricken sometime in the future. All humans have two copies of each chromosome and

therefore of each gene, one from the mother, one from the father. Some disease states are not caused by having a single copy of the affecting gene, even for single-gene diseases, but require that both copies of the gene be defective to be manifested. Persons carrying one copy of such a defective gene are carriers of a potential disorder for their offspring, but are not affected themselves.

These additional complexities of genetic information can lead to some situations seldom encountered in considering the privacy and discrimination issues raised by other types of medical data.

The Ethical, Legal and Social Implications (ELSI) Program

Recognizing the potential for misuse of the increased genetic knowledge that will become available in the future, DOE and NIH have set aside approximately 3 percent of their human genome budgets for programs focused on the critical policy and legal issues as well as on public awareness of both the basic scientific facts and the societal issues concerning genetic knowledge. The educational component of the program is designed to increase public awareness of the basic facts of genetics, the capabilities of DNA technology and the related issues that concern the public. A basic level of understanding of the scientific facts by the public is a prerequisite for intelligent involvement in a discussion of the societal issues.

DOE's program was initiated slightly over a year ago. Since our projects have only been funded within the past six months, I cannot report any conclusions at this time. I will, however, describe ongoing activities and planned

efforts for the future to give the Subcommittee a view of the spectrum of issues and approaches being taken. The areas of emphasis in the respective DOE and NIH programs are distinct although they remain complementary and the agencies will continue to cooperate closely. In fact, DOE and NIH co-sponsor a Working Group of outside consultants chaired by Dr. Nancy Wexler, that helps to coordinate the two components of the program. Dr. Wexler will describe the operation of the Working Group in her testimony.

In the immediate future, DOE will emphasize studies and educational activities related to the issue of genetic privacy, although we will not exclude support of worthy projects in the other ELSI areas. Activities sponsored by DOE include empirical and analytic studies of the issues from the perspectives of law, medicine, the humanities, the social sciences, and public policy. The program also supports conferences of scholars and other involved and interested persons aimed at exchanging views and presenting the results of analyses. The educational activities sponsored by the program include the production of television documentaries and the preparation and distribution of high school teaching materials.

More specifically, the DOE ELSI program currently supports two research efforts, two conferences, three educational projects, and several smaller projects. The research efforts include surveys of legislation on genetic privacy and the views of insurers and insurance regulators on the use and abuse of genetic information. We will also co-sponsor with NIH a study by the Institute of Medicine entitled, "Predicting Future Disease: Issues in the Development, Application and Use of Tests for Genetic Disorders." In

addition, two conferences will focus, respectively, on genetic discrimination and a theological perspective on the ELSI issues. The educational projects include: a documentary series on "Medicine at the Crossroads," co-sponsored with several federal agencies, the preparation and distribution to high school biology teachers in the United States of a teaching module on the Human Genome Project and the ELSI issues, and a workshop for persons in science communications and administration on the theory, practice, applications and implications of human gene manipulation.

During the past week, DOE conducted its annual peer review of applications for ELSI grants during the coming year. This round, DOE received over three times the number of applications of the previous round. I would like to submit to the Subcommittee a descriptive list of the projects currently supported by DOE as a more detailed view of the nature of the program.

Genetic Privacy and Genetic Discrimination: Two Sides of the Policy Coin

The opportunities for the misuse of genetic data on individuals will increase with time, as genetics comes to play an increasingly prominent role in medicine. Consequently, the DOE-NIH ELSI programs have identified "privacy" as one of the most crucial ethical, legal and social issues concerning the knowledge to be gained from the Human Genome Project. Indeed, all three areas of concern -- the ethical, legal, and social -- are affected by privacy.

While privacy is of recognized importance, it is clear that the other side of the coin from a policy point of view is the issue of discrimination - what happens when information does become public. It is clear that the question

here for rule-makers and law-makers, in its simplest form, is what right and obligations, if any, should employers, insurance companies, medical care organizations or any of the potential users of genetic information, have to discriminate or not discriminate among individuals on the basis of their genetic make-up?

Although we are at an early stage in our efforts, I would like to point to some issues that have been raised in program-sponsored meetings concerning privacy protection in the area of genetics. We do not have solutions. Indeed, our principal objective now is to inform the discussion in the ethical, legal and social areas. From this discussion, approaches to solutions may emerge. The first steps have been taken, however, by posing some of the important questions.

These questions include the following. What is the real nature of current limits on existing legal protections of genetic data? For genetic information, what are the limits of concerns for public health and safety when weighed against the individual right to privacy? How is the privacy of one individual's genetic information affected by the rights of another, genetically related individual? How well is genetic data actually protected in practice? How do other countries protect privacy? What social effects of their policies are evident, if any? Does the character of the country's health-care system affect the privacy or discrimination issue? Do foreign laws and practices provide models for privacy protection in this country?

These issues, as well as the issues mentioned above related to potential public uses of genetic information are certainly not simple.

Conclusion

The Human Genome Program is already producing new technologies and a vast amount of genetic data that will catalyze new biological insights into the functioning of the human body. It will light the way for all of us to understand better why we are the way we are, and will provide tools for medicine that, with other ongoing biomedical research, will constitute a real revolution. The prospect of new understanding must make us reflect on its impact on society. The new information raises few really new issues, but the relevant old issues have not been adequately dealt with - that is the challenge. We must now engage some difficult questions of public policy in order that the benefits of the revolution in science and technology will not be overshadowed by the darker side of human behavior in discrimination and misuse of the precious fruits of our science. I hope that the information and views I have provided will prove useful to the committee in their future considerations of these very important issues.

That concludes my statement. I will be happy to answer your questions.

Statement of

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Joint NIH/DOE Working Group on the

Ethical, Legal and Social Issues

of the Human Genome Project

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Hereditary Disease Foundation

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before the

United States House of Representatives

Committee on Government Operations

Subcommittee on Government Information, Justice and Agriculture

October 17, 1991

INTRODUCTION

I am honored to be here this morning. You are to be congratulated for focussing the nation's attention on matters fundamental to each of us as individuals and as citizens, that is, protection of the privacy of medical and, particularly, genetic information. I would like to talk with you today about the rights of persons for the maintenance of privacy of genetic information. But I would also like to speak to the essential rights of persons to disclose genetic and medical information with impunity, without fear of harmful reprisals. And I would like to address the rights of individuals to choose whether or not to preserve their privacy or speak openly without negative repercussions.

I have the privilege of serving as Chairperson of the Joint NIH/DOE Working Group on the Ethical, Legal and Social Issues associated with mapping and sequencing the human genome. I am also speaking to you today as the President of the Hereditary Disease Foundation, and as Associate Professor of Clinical Neuropsychology in the departments of neurology and psychiatry, College of Physicians and Surgeons, Columbia University.

I have had personal experiences with respect to issues of genetic privacy and disclosure which I think are relevant. My mother was diagnosed with Huntington's disease when I was 22 years old. The illness had already claimed the lives of my maternal grandfather and three uncles. My father explained to my

older sister and me that we each had a 50/50 risk of inheriting it. The disease causes uncontrollable movements in all parts of the body, intellectual deterioration and severe emotional disturbances. It is invariably fatal and essentially untreatable.

I went to graduate school immediately after learning of this upheaval in our lives. Although I had done nothing to bring this on, I was at first embarrassed and ashamed to tell anyone about my mother's sad decline or my own risk. I was afraid people would treat me differently, watch me for symptoms, not want to date me, be overly distant or too solicitous. In graduate school I become involved with working with families with Huntington's disease but for some time I kept that world and my academic life quite separate. It felt slightly schizophrenic, literally commuting between my world of families with Huntington's disease in Detroit and my academic life in Ann Arbor, Michigan. Thanks to exceptionally understanding and wise faculty and friends at the University of Michigan, my two worlds meshed in very gratifying ways. But when I applied for an academic position after completing my doctorate, I asked all my advisors to rewrite their letters of recommendation because, although they had said nothing explicitly, I was afraid that it would be too obvious why I was interested in Huntington's disease. I was very concerned that I would never be hired if my risk status was known, and certainly never be considered for tenure.

The turning point came for me when the National Institute of

Neurological Disorders and Stroke invited me to serve as Executive Director of the Congressional Commission for the Control of Huntington's Disease and Its Consequences and later to join their staff. For the first time I could be totally open about the disease without fear of alienating my colleagues or losing my job security and employment benefits. The Neurology Institute was even courageous enough to offer me a civil service job from which it is difficult to extract people. This experience was enormously healing because my colleagues, experts in the disease, were willing to take a chance on a person with a one in two possibility of developing a neurodegenerative disease of the brain and body, with insidious onset, causing failures of judgment and memory and emotional instability. It was not privacy in this instance that was necessary - it was candor that cured.

The leadership of the Human Genome Project, both at the NIH and the DOE, were also willing to place a representative of genetic "consumer" groups as chair of their joint Ethical, Legal, and Social Issues Working Group. While many in genetic support groups have trepidations regarding the utilization of genetic information, the fact that the NIH and DOE have launched the biggest biomedical ethics program nationwide concomitantly with support of the basic research has helped to allay peoples' concerns. Much needs to be done, but at least there are resources available to do it thoughtfully, with careful planning, and there is interest at the NIH, DOE, and most importantly, on

Capitol Hill.

Let me emphasize that although no formal poll has been taken, the vast majority of families with genetic disorders are enormously grateful for advancing genetic technology. To us, it represents the best hope for an effective treatment and even eventual cure. Some in government or the general public have suggested slowing down the technology until the social support systems "catch up." This is antithetical to the best interests of patients and families as technology is the only hope that many have who are in a race against time before the effects of a lethal gene overtake them. When treatments exist, there will be fewer incentives to discriminate and the burden of revelation will diminish.

THE PATHWAY BETWEEN PREDICTION AND PREVENTION

It is important to understand the stages and time course by which most research on genetic disease progresses. First DNA markers must be identified which are localized precisely along a chromosome so that each is a small and specific distance from the other, like distance markers along a highway. Then, by studying families with a particular genetic disease and watching the disease gene being passed from generation to generation together with certain markers, the disease gene is "mapped" to a specific chromosome. Once close markers are discovered, presymptomatic and prenatal diagnosis is possible -- even decades before the

appearance of the illness. But the aim of the research is primarily therapeutic, not just diagnostic. Early diagnosis can be a benefit in many instances in which early intervention is critical, such as cancer or heart disease, or problematic if there is no prevention possible, such as for familial Alzheimer's disease.

The next step is the isolation and characterization of the abnormal gene itself. This phase can be rather rapid, a year or two, or can take a very long time. The HD gene was localized in 1983, the first instance in which DNA markers were successfully used to map a gene whose chromosomal assignment was unknown. Eight years later, we are still searching for the gene. The Human Genome Project will dramatically shorten the time it takes to find and characterize genes; at the successful conclusion of the Project one will only need to look it up in a book.

Observing altered genes and studying homologous genes in plants and animals can give rise to suggestions for therapy. Scientists are now exploring novel delivery systems using inhalants such as those used for treating asthma to place normal genes in the lungs in order to treat cystic fibrosis or alpha-1 antitrypsin disease. Some scientists are capitalizing on the affection which cold viruses, adenoviruses, show for the lung to harness them into the service of transporting normal genes in to correct damaged lungs.

The Human Genome Project greatly expedites finding and characterizing normal and abnormal genes in human and model

organisms and develops new technology that facilitates gene therapy. Gene therapy cannot be approached until the offending gene is identified and understood. Although understanding the molecular lesion is no guarantee of a cure, a sad lesson learned from sickle cell research, many new avenues are surely opened.

Another advantage of the Human Genome Project is that once critical genes modulating important biochemical or cellular pathways are discovered, they may be found to play a role in both the hereditary and sporadic forms of an illness. For example, both familial and sporadic forms of Alzheimer's disease may involve a disruption of the beta-amyloid gene. Treatment for the much more common sporadic form may be the same as treatment for the familial variant. Neurofibromatosis, retinoblastoma, colon and breast cancer all may involve disruptions of oncogenes, which produce tumors, or tumor suppressor genes. An environmental insult may work its effect by disrupting the action of these same genes in a cell, affecting somatic genes rather than germ cell genes. Although the disorder produced is not <u>hereditary</u> in that it is not passed on to the next generation, it is still <u>genetic</u> in that the functioning of a single or possibly multiple genes are affected. New treatments for cancer, for example, may address correcting the damaged gene, even though a noxious chemical might be the instigating agent of the disease.

Even some people's differential response to the AID's virus has been shown to be genetically mediated. Learning how these people resist the illness may lead to new therapeutic

understanding. Transplant rejection occurs due to genetic incompatibility between host and donor; if the actions of those genes could be modified, transplant success rates could improve dramatically. Of the approximately 100,000 genes that humans possess, probably the vast majority exist in some altered state through the effects of mutations through the millennia and these disordered states contribute to today's morbidity and mortality. The Human Genome Project should set the stage for the development of extensive new therapies both for disorders known to be hereditary and even those that are not. The benefits to human health are incalculable.

The risks are to some extent known. Dangers posed by genetic knowledge challenge systems already strained in our society today. These problems preexisted the Human Genome Project and they are too extensive to be ameliorated by our efforts alone. But the Human Genome Project can contribute toward their solution by convening and supporting some of the brightest and most creative thinkers to focus their efforts on devising policy and program recommendations.

THE JOINT NIH/DOE ETHICAL, LEGAL AND SOCIAL ISSUES WORKING GROUP

The mandate of the Joint NIH/DOE Ethical, Legal and Social Issues Working Group of the Human Genome Project is to anticipate problems attendant on this burgeoning technology and make programmatic and policy recommendations to assure that

information is used for the benefit of individuals and society. The National Center for Human Genome Research, National Institutes of Health, program on Ethical, Legal and Social Implications (ELSI) is directed by Dr. Eric Juengst, a philosopher trained in ethics with experience both in genetics and in the humanities. The ELSI program of the Department of Energy is directed by Mr. Michael Yesley, a lawyer who served as staff director of the National Commission for the Protection of Human Subjects in Biomedical and Behavioral Research and who has long-standing experience with issues of privacy and confidentiality. The joint NIH/DOE ELSI Working Group serves in an advisory capacity to the Human Genome Programs of both parent institutions. It is comprised of experts in medical genetics, ethics, law, biology, psychology and sociology. In each of its quarterly meetings focussing on different topics of interest, additional experts in diverse areas are convened.

The first priority of the Working Group is to facilitate the distribution of grant funds to support investigators and policy makers in the larger community. The testimonies of Dr. Bernadine Healey, Director of the NIH, and Dr. David Galas, Associate Director for Health and Environmental Research, DOE, cover the details of these programs. Certain areas are of such high priority that the Working Group itself has taken the initiative to develop programs in these areas. The ELSI Working Group enunciated four areas in which immediate attention is required:

1.) research on issues of quality and access in the use of genetic tests

2.) research on the fair use of genetic information by employers and insurers

3.) research on privacy issues involving genetic information

4.) public and professional education

REPERCUSSIONS OF DISCLOSURE -- INSURANCE COVERAGE

In the United States, with our current system of insurance coverage, unwanted disclosure of genetic information can result in the loss of critical health benefits, not only for an individual but for his or her entire family. It would be a bitter irony if people who can benefit from early diagnostic tests are dissuaded from availing themselves of the test because they may loose the very insurance they need to prevent the disease or protect themselves from it when it appears.

The medical community and the public were elated recently when two scientific groups announced the discovery of a gene causing polyposis coli or colon cancer. The early diagnosis of colon cancer can be life saving. If persons who carry one copy of this autosomal dominant gene have a colectomy before symptoms start or remove colon adenomas which are the first signs of developing disease, they can very effectively prevent colon cancer. If they cannot afford to pay for the genetic test, if third party carriers refuse to reimburse for it, or delete

coverage of the necessary preventive procedures, the advantages of early diagnosis will be for naught. The families who participated in research that led to these breakthroughs in gene identification, families in whom it is already known that this will be an effective and accurate test, are now cautiously weighing their options.

If you think that this is a fictitious concern, consider the following. A woman in Michigan had two siblings affected by colon cancer. She wisely had a colonoscopy as a prophylactic measure the results of which proved to be perfectly normal. She later applied for a health insurance policy. Although neither she nor any of her physicians discussed the family history of colon cancer with the insurance company, they somehow acquired this information. When her new policy arrived it contained a rider excluding all coverage of procedures relating to her colon. If she should develop colon cancer in the future, she would have to assume all treatment costs herself -- an impossibility. She cannot even afford to pay for the colonoscopies which might minimize the cost of any cancer through early detection.

Although her own genetic concerns should have been sufficient concern for one individual, this same woman married a minister who later developed Huntington's disease. The fatal HD gene was passed on to both their children who manifested the illness at a young age. Huntington's disease has a gradual onset. The neurologist diagnosing the illness in the son waited until he was age 19 to spare him the devastating news. What

doctor and mother did not realize was that had the son been diagnosed at age 18, when he was clearly symptomatic, he would have been covered as a dependent of the mother's on her insurance policy. As it was, he was independent but already ill and totally uninsurable. The mother's policy, although it had extensive mental health benefits for family members, was useless to her because Huntington's disease was defined as requiring custodial care and no amount of insurance would cover such care. This is often the situation confronting families with hereditary diseases that are chronic in nature. The mother was faced with the prospect of state hospital placement for her husband and son, both of whom were totally dependent on her modest earnings for their support.

The Insurance Task Force of the joint NIH/DOE Ethical, Legal and Social Issues Working Group is addressing critical questions with respect to how the insurance industry responds to the introduction of new genetic tests. Their recommendations, due in 1993, should help shape policy and practices in these areas, rather than merely being reactive to industry stances. (The Task Force is focussing on health insurance alone. Most people with or at risk for hereditary disorders are ineligible for life insurance.) As more new tests are developed, both for illnesses considered to be strictly inherited and for those in which some cascade of genes may play a role, public awareness of insurance industry practices will be heightened. An increasing number of individuals who took their insurance coverage for granted may

find themselves among the group whose insurance is in jeopardy.

At the moment, some individuals taking genetic tests are paying for them out-of-pocket to avoid any potential insurance repercussions. Presymptomatic testing for Huntington's disease is a "linkage analysis," studying DNA markers "linked" or very close to the HD gene to make a diagnosis. It is more accurate with more persons in the family tested using more markers. Some persons are paying up to $4,000 for testing; the expenditure is worth it to them to keep the information private. It is unclear if third party carriers would pay for the test and what actions they would take following a diagnosis of either positive or negative for the gene.

The insurance companies, in turn, are concerned that the public will pay personally for genetic tests and then alter their insurance coverage according to the outcome: increased insurance if health problems are predicted, decreased insurance for those shown to be healthy. This "adverse selection" might skew actuarial calculations in a deleterious fashion.

Some insurance companies are beginning to propose new programs for pooling risks across a large number of small employers and providing for more universal coverage, regardless of risk. They recognize that according to their current criteria, a large segment of the American public may become uninsurable with the advent of new genetic tests. Certainly the aim of the Human Genome Project is not to swell the ranks of the 37 million uninsured in this country. If, however, under the

auspices of the Human Genome Project representatives of the insurance industry, of consumer groups, academia, government and the public can meet to develop new policies and programs we can help to catalyze constructive changes for a much larger constituency.

REPERCUSSIONS OF DISCLOSURE -- EMPLOYMENT

Issues of insurance are integrally enmeshed with fears regarding employment discrimination. With health care costs soaring, there is a strong economic incentive for employers to screen out individuals who will develop costly illnesses. In some instances, the employee may be perfectly healthy but be a carrier of a deleterious gene that might combine with the same defective gene in a spouse to produce a devastating disease in offspring.

A recent report by the Office of Technology Assessment indicated that 12 companies among the Fortune 500 and 50 major utility companies reported using any form of genetic screening or monitoring in the workplane. Although the number is small, it may be because the full ramifications of genetic testing and its consequences have not seeped into public awareness sufficiently to entice employers. The attitude of insurance carriers toward genetic testing may help shape those of employers.

The recently passed Americans With Disabilities Act (ADA) forbids discriminating against qualified individuals with a

disability with respect to job application procedures; hiring, advancement and discharge practices; and compensation. The Act provides extremely important protections against employment discrimination for those either disabled by, presymptomatically identified to have, or asymptomatic carriers of a genetic disease. Taking its cue from the seminal Vocational Rehabilitation Act of 1974, the ADA protects three groups of individuals defined as disabled. The first definition, persons with a physical or mental handicap that substantially limits major life activities, will protect persons currently disabled due to a genetic illness. The second definition, those with a history of such an impairment, should protect persons with a treatable hereditary disease who may have been incapacitated but are now quite functional, such as persons with PKU, Gaucher's disease or hemochromatosis. The third definition of disability protects a group of people new to our citizenry but growing: the presymptomatic individual. The third definition includes persons regarded to have a such an impairment. This third definition should protect me against employment discrimination if an employer chooses not to hire me only because I am at risk. If I took a presymptomatic test for Huntington's disease and was shown to most likely have the gene (the test is not 100% accurate yet), my employer and I would know that the disease will at some point appear. And yet, as I am still functional and asymptomatic, my employment assessment should focus only on whether or not I can

do the job, not on the fact that some day I will no longer be able to function.

The ADA, and the EEOC interpretations of it as set forth in their regulations, specify that employers may only use job-related medical criteria in hiring decisions. Under the EEOC regulations, employers are permitted to perform a variety of medical tests once an offer of employment has been made, conditional on the outcome of a medical examination. Although employers are not legally restricted in the tests they request, they are only entitled to use job-related medical information.

The Joint ELSI Working Group and the Chairmen of the NIH and DOE Genome Advisory Committees wrote to Mr. Kemp, Chairman of the EEOC, requesting that permissable medical examinations be limited to assessing only job-related physical and mental conditions. We felt that even though employers were only entitled to utilize job-related medical information in their hiring practices and even though employment offers contingent on medical information must depend only on job-related medical results, permitting employers to perform any and all tests encourages surreptitious testing. If an employer legally cannot utilize certain medical information, why permit the employer to gather that information? Why would an employer pay to perform non-job-related medical testing, not at the voluntary request of the applicant or employee, if the information cannot be legally utilized in hiring decisions? To what use will that information be put?

The letter from the NIH/DOE ELSI program to the EEOC also

recommended that the ADA specify that unaffected individuals who are heterozygous carriers for a gene causing a recessive (or X-linked) disease, that is carriers of one copy of a gene which, when two copies are present, causes disease in offspring, be explicitly protected under the Act. An example would be a person carrying one gene for sickle cell disease or cystic fibrosis. The carrier is asymptomatic. An employer may be tempted to discriminate, however, because if a carrier employee has a child with another carrier, each offspring has a one in four chance of having an expensive disease. (The annual cost of medical care for a patient with CF is about $20,000. Lifetime medical costs, costs based on a median lifespan of 27 years, are approximately $500,000.) The carrier rate for cystic fibrosis among Anglo Saxons is 1 in 25, while 8% of Afro-Americans carry the sickle cell gene. We are speaking of common disorders.

The ELSI program also recommended that the ADA deal with the privacy of genetic and medical information as a way to protect employees against discrimination. When insurance claims are made, usually an entire chart arrives in the company benefits office for other employees to peruse easily. Medical records are not "sanitized" so that only relevant material travels to other medical referrals or to benefits offices. We were informed by staff of the EEOC that our recommendation with respect to privacy exceeded the scope of the ADA.

THE PRIVACY AGENDA

The ELSI Working Group has established a Privacy Task Force under the direction of ELSI member Ms. Patricia King, professor of law at Georgetown University, who has a long and distinguished career of government service as a member of the National Commission for the Protection of Human Subjects in Biomedical and Behavioral Research, and the National Institutes of Health Panel on Human Fetal Tissue Transplantation Research, among many positions. The Privacy Task Force is just being organized and setting its agenda. Among the areas in which research and policy recommendations are sought are the following:

Genetic Services

The ELSI supported Institute of Medicine study on "Predicting Future Disease: Issues in the Development, Application and Use of Tests for Genetic Disorders" will also focus attention on these most crucial questions of privacy and disclosure in the provision of genetic services. Genetic information, unlike most other medical information, immediately reveals private and personal information about others connected to the individual in question. For example, if you learn that my mother had Huntington's disease, you immediately know something intensely personal about my sister and me without our saying anything about ourselves. When I took my mother to a new doctor one day, he said to me, "Oh, Huntington's -- you have a one in

two chance of having it too, no?" He had no idea what I knew or didn't know.

There are controversies brewing within families which may spill over into courts with respect to ownership of genetic information. In certain genetic disorders for which close markers have been found but the gene not yet isolated, linkage tests using these closely linked DNA markers are the only means of providing diagnostic information. DNA samples from specified relatives are required. Linkage tests are now being used for presymptomatic diagnosis of Huntington's disease and polycystic kidney disease, among others. In some instances, parents have refused to give a blood sample for a test on the grounds that the counseling provided at a certain center was inadequate. In other instances, parents wished to provide genetic information for one offspring but not another. Once the information was given for the first, however, it was already known for the second. Could the information be used without a parent's permission to honor the request of a person at risk to learn his or her own genotype or should the request for the privacy of genetic information on the part of the parent be honored? The privacy of one person can be detrimental to the autonomy of another.

Another instance in which privacy may be violated is when a physician determines that there is a serious danger to others based on their relationship to someone with a known genetic problem. Not infrequently, the genetically affected individual requests that no one know the news, including prospective

marriage partners or siblings and cousins who may be equally at risk. The client is too ashamed or embarrassed, frightened or distraught by the information to inform others or even to allow the physician or genetic counselor to do so. The President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research in their Report on Screening and Counseling for Genetic Conditions discussed criteria for determining when patient confidentiality should be honored or abridged. Although complying with clients' desires for privacy, the report enumerated circumstances in which geneticists could responsibly contact relatives or others whom they deemed needed to know genetic information without permission of the client. This can occur when all efforts at persuasion of the initial client fail and if the condition in question poses a serious harm, such as an unbalanced chromosomal translocation with a high likelihood of resulting in severe disabilities in any child born with the genetic problem.

This problem of "contact tracing" to borrow a phrase from the public health model of infectious disease control, is likely to become more prominent as additional genetic tests are introduced. If about one in 25 caucasians carries an abnormal gene causing cystic fibrosis, once one family member is identified there will be an incentive to find the rest. But some individuals may prefer not to know if they are carriers and their wish for privacy must be respected.

Knowing and Not Knowing: Co-equal Rights?

Western culture is one in which knowledge is valued highly -- "knowledge is power," and to avoid knowledge is looked upon pejoratively -- "to hide your head in the sand". And yet, new genetic knowledge has enormous repercussions for individuals and families. In some instances, predictive testing exists for diseases for which there is no treatment or cure, such as Huntington's disease or neurofibromatosis. We can tell people that they will surely die of HD but we cannot tell them when the disease will appear. We can tell people that their children will have neurofibromatosis but we cannot tell them how severely the children will be affected, ranging from a few large "freckle"-like patches called cafe-au-lait spots to numerous disfiguring tumors all over the body.

For many who are at risk for untreatable late onset diseases, such as familial Alzheimer's disease or amyotrophic lateral sclerosis (ALS or Lou Gehrig's disease), the availability of presymptomatic testing forces them to contend with issues of timing. Should they know prematurely, before the disease begins or wait until symptoms start? Do they have the luxury of choosing for themselves or are there instances in which others require this knowledge? What if you were to know for certain that I am going to develop Huntington's disease. Would my university be reluctant to hire or promote me? Would Drs. Watson and Galas have entrusted this important committee to my leadership? If I were training to be a neurosurgeon, would you

feel it your ethical duty to persuade me to pursue a profession in which coordination, dexterity and judgment, lost early in HD, were not at a premium? Are there instances in which employers have a right to know genotypes or should they judge only on proficiency? If you knew for certain that I was going to develop Huntington's disease, would you feel any differently toward me now?

Testing Minors

Privacy issues can be particularly complex when one party is not able to provide informed consent to the abrogation of privacy. The current policy of testing centers for Huntington's disease, and probably for other late onset disorders as tests are developed, is not to test minors who are unable to provide informed consent for themselves. This policy defies common practices described by family law in which parents are entitled to medical information with respect to their minor children.

The clinicians and families developing the Huntington's disease testing protocol felt it was too onerous a burden for children to carry, knowing they are destined to die of Huntington's disease, and there is no medical advantage to knowing this information early. Testing centers also have refused to test children awaiting adoption, either at the request of prospective adoptive parents or the agency. One day, however, parents who feel justified in invading the privacy of their children for planning purposes may bring the issue to court.

Research on Large Families

One of the first activities of the ELSI Privacy Task Force will be to focus on the protection of privacy in the conduct of genetic research. Now that investigators are successfully localizing genes, they are faced with questions regarding recontacting individuals within the families participating in the research to inform them of their genotype, if they request it. Much of the confusion over recontacting could be alleviated by discussing the issue of providing feedback before the research begins, even specifying in informed consent forms whether and how such recontacting should take place. Some individuals will participate in the research only under conditions of anonymity and do not want to know their genotypes; others explicitly desire to know. Both should be accommodated.

In France recently, an interesting conflict occurred between a scientific group that had acquired information, through geneological studies, regarding which children in a small region were particularly at high risk for developing hereditary juvenile glaucoma. This genetic condition must be treated early or it will result in permanent blindness. The investigator's plan to contact high risk families immediately collided with French privacy law which forbade them to do so. The issue was finally resolved by the scientists mounting an intensive educational campaign in the relevant region, warning parents of the dangers and encouraging them to contact their local physicians who had

also been briefed by the researchers with all information except identifying names.

The frustration on the part of the investigator is to have information critical to someone's health, if early intervention is required, and be prevented from directly contacting the person. On the other hand, if the intervention is only marginal and the information devastating, individuals may not welcome such intrusions on their privacy.

Another area of concern is how research results are published. Following the tradition of accuracy in publishing, some groups have found themselves in the painful position of publishing genetically revealing information in the correct pedigree form so that it is recognizable to family members; should they ever see the publication, they will learn of a death sentence for certain relatives. And scientists should assume that eventually research subjects will see all relevent publications. Other groups disguise pedigree information, indicating that it is altered. Uniform standards for publishing sensitive information must be developed.

Data Bases and Banks

An important focus of attention for the Privacy Task Force is on banks and data bases either containing blood samples, DNA, or sensitive information pertaining to genotype. These banks range from the immense DNA banks being established for forensic purposes, to commercial banks, to academic and research banks, to

military banks, to sperm and ova banks, to public health banks, to "bottem drawer banks" of individual physicians or investigators, to voluntary health associations, to schools, to companies -- the list is expanding on a daily basis. With the advent of PCR techniques, spots of DNA used for testing babies at birth for a variety of metabolic disorders can now be re-utilized to check for hereditary diseases for which the parents never originally gave consent. There is growing discussion over how long samples should be kept, whether they be blood samples collected at birth or DNA samples of convicted felons. A critical issue is maintenance of security of the data and samples, particularly for interstate and international collections.

There is also potential controversy brewing over the use of DNA samples collected for one purpose and used for other, unrelated purposes. For example, if genes purportedly predisposing toward aggressivity, explosive discontrol syndromes or sociopathy were discovered, should investigators be permitted to search for these genes in DNA samples extracted anonymously from felons' DNA banks? The Center for Disease Control is now collecting transformed lymphocyte lines as part of its HAINES study examining general nutritional and population variables. To what use should these samples be put? We need to be very imaginative in our conjectures regarding the use of genetic materials: President Lincoln surely never suspected that his bones would be screened for the presence of a gene causing

Marfan's syndrome.

THE MISUSE OF GENETIC INFORMATION

Racism Under the Sheet of Genetics

Sociologist Troy Duster, in his recent book Backdoor to Eugenics, expressed the concern that since different ethnic groups are differentially affected by certain genetic disorders, racial prejudice will reemerge with renewed strength under the guise of genetic interest. One is not discriminating against Afro-Americans, an illegal activity, only people who are carriers of the sickle cell trait, who just happen to be predominantly Afro-American.

Screening for sickle cell anemia in the 1970s demonstrated how a program that was initially thought to be beneficial to a population resulted in doing harm. There was tremendous public confusion over the difference between those who were symptomatic and asymptomatic individuals with only a single copy of the sickle cell gene. Both patients and carriers lost their jobs, their insurance and suffered greatly. Even the U.S. military misunderstood the consequences of being only a carrier.

Genetic Education

Sensitive genetic information is entering a climate that is not very much more sophisticated today. There is a dearth of trained genetic professionals: fewer than 1,500 medical

geneticists and genetic counselors in this country. Many physicians have an inadequate understanding of genetics due to the paucity of genetic training offered in medical schools. Not only must complex notions of probability be communicated to clients requesting genetic information, but the great variability of genetic illness must be explained.

The ELSI programs of NIH and DOE are addressing these problems through the IOM study of genetic services and support of the development of new school curricula. Public awareness is being sharpened through support of two PBS television programs under development. Additional new incentives and programs will be a high priority for ELSI attention.

The Nazi Era

Dorothy Nelkin and Lawrence Tancredi, in their book Dangerous Diagnostics, discuss the possibility of developing a new "biological underclass" of people unable to obtain employment or insurance benefits, discriminated against in an increasingly "medicalized society". The authors present a possible worst case as a way of preparing us to provide the necessary protections against this outcome.

Nazi Germany has already provided us with a horrific example of the depths to which humans are capable of descending. (For information in the following sections, I am indebted to Dr. Peter Harper, editor, Huntington's Disease, WB. Saunders Company Ltd., London, 1991, pg.365-369.) The racial hygiene policies of the

Third Reich selected two medical groups for sterilization or elimination: the mentally ill and handicapped and those affected by genetic disorders. As persons with Huntington's disease fit both categories, they were doubly jeopardized. Nine disorders were listed for which sterilization was mandatory in the compulsory sterilization law of July 14, 1933: hereditary feeble-mindedness, schizophrenia, manic-depression, hereditary epilepsy, Huntington's chorea, hereditary blindness, hereditary deafness, hereditary malformations, and severe alcoholism. Patients to be sterilized were brought before a "Genetic Health Court" which ruled on diagnosis and recommended for or against sterilization.

Friedrich Panse, who died in 1972, was a professor at the Psychiatric-Neurological Research Institute in Bonn, whose director was Professor Kurt Pohlisch. Both were Nazi party members instrumental in establishing the race-hygiene laws and were actively involved in the genetic health courts and mass murder. Both were acquitted after the war and returned to prominent positions in their universities. Panse conducted the first survey of Huntington's disease in Germany, amassing a great volume of family record data. He also reported all cases and their families to the Nazi health administration where they were later sterilized or murdered.

There is currently a tremendous controversy in Germany regarding the use of Panse's register which has been recovered following the integration of east and west Germany. There is currently a moratorium on the use of these materials while debate

rages as to whether or not records gathered by a Nazi official for sterilization or elimination purposes can serve a legitimate research or medical function.

There were 350,000 to 400,000 people compulsorily sterilized during the Nazi regime. Beno Muller-Hill estimates that there would have been about 3,000 to 3,500 sterilizations for HD. When sterilizations no longer sufficed, mass murder ensued and Huntington's disease was on the list specifying disorders for which extermination was required. The numbers killed is unknown.

Local Politics

Although these atrocities occurred in Germany, the U.S. has nothing to be smug about. Charles Davenport, a prominent eugenicist who headed the Cold Spring Harbor laboratory before Dr. Watson's time, supported the German program of compulsory sterilization and recommended a similar policy at home:

> It would be a work of far-seeing philanthropy to sterilize all those in which chronic chorea has already developed and to secure that such of their offspring as show prematurely its symptoms shall not reproduce. It is for the state to investigate every case of Huntington's chorea that appears and to concern itself with all of the progeny of such. That is the least the state can do to fulfil its duty toward the yet unborn. A state that knows who are its choreic and knows that half of the children of every one of such will (on the average) become choreic and does not do the obvious thing to prevent the spread of this dire inheritable disease is impotent, stupid and blind and invites disaster. We think only of personal liberty and forget the rights and liberties of the unborn of whom the state is the sole protector. Unfortunate the nation when the state declines to fulfill this duty! (Davenport and Muncey, 1916).

CONCLUSIONS

We meet here today with the nation on a very different mission than that prescribed by Davenport. We meet to protect the privacy of individuals and families, to prevent the abuse of genetic information or the loss of rights and liberties.

Some people point to the Nazi catastrophe as evidence of the dangers of genetic information. But this barbarism occurred a decade before James Watson and Francis Crick discovered the structure of DNA, long before genes were localized on chromosomes or we were capable of sequencing them. The Nazis relied on observable symptoms, violations of the privacy of an individual emanating from the genes themselves.

It is chilling for me to see my name, so to speak, on the Nazi list slated for extermination or sterilization or read Davenport's cruel words. But it was my own torment to watch my mother in her personal concentration camp enslaved by a mind and body that no longer functioned. One in four persons with Huntington's disease attempts suicide and my mother was among this group. The Nazi activities were barbaric beyond imagination. But many hereditary disorders are also barbaric for those that suffer from them and those that love them. We cannot allow our concerns about the potential misuse of genetic information retard the search for alleviating the physical and psychological pain of these illnesses.

In summary:

1.) The Human Genome Project is an egalitarian search for all genes, many of which can cause or contribute to causing genetic disease.

2.) The Human Genome Project is an organized, coordinated and collaborative effort to find genes. The majority of Voluntary Health Organizations cannot support the piecemeal, expensive endeavors that may be required to find "their gene".

3.) The joint NIH/DOE Working Group on Ethical, Legal and Social Issues intends to be proactive and energetic in helping define a social and medical agenda in which people can take advantage of the benefits of new genetic knowledge without suffering from discrimination, economic, social or psychological loss, or stigmatization.

4.) New legislation may be required, either on a Federal or state level, to ensure the privacy of genetic information. ELSI programs will investigate the advantages and disadvantages of state versus federal legislation and help to develop policy options for introduction into new legislation.

5.) Discrimination based on genotype, just like discrimination based on race or gender which are expressions of genotype, should be prohibited. Legislation may be required to reinforce this basic civil right.

6.) The fruits of the Human Genome Project are a source of great hope for millions of Americans. When I ask people who are presymptomatically diagnosed with Huntington's disease what

sustains them, they usually answer "God and trust in science." Our political agenda is complex and will demand empathy, caution and courage. It cannot be carried out at the expense of science, but as two complementary programs to prepare for the 21st century.

ONE HUNDRED SECOND CONGRESS

Congress of the United States

House of Representatives

GOVERNMENT INFORMATION, JUSTICE, AND AGRICULTURE
SUBCOMMITTEE
OF THE
COMMITTEE ON GOVERNMENT OPERATIONS
B-349-C RAYBURN HOUSE OFFICE BUILDING
WASHINGTON, DC 20515

October 9, 1991

Dr. Paul Billings
Department of Medicine
California Pacific Medical Center
P.O. Box 7999
San Francisco, CA 94115

Dear Dr. Billings:

On Thursday, October 17, 1991, the Subcommittee will continue a series of hearings on domestic and international data protection issues. The purpose of these hearings is to explore data protection problems faced by government, consumers, and businesses, and to identify solutions to those problems. The first hearing in this series was on public and corporate reaction to privacy. It covered a recent public opinion poll on privacy and the development and demise of Lotus Marketplace.

The subject of the upcoming hearing is possible uses and misuses of genetic information. The Subcommittee will consider the changing nature of genetic information, the ways in which the information can and should be used, the effects on the individual of misuse, and the role of the government in establishing rules for the collection, maintenance, disclosure and use. The Subcommittee's interests include the federal government's use of genetic information as well as how the information can be used or misused in education, insurance, employment, and other ways.

I invite you to testify at this hearing. The hearing will begin at 9:30 a.m. in Room 2247 of the Rayburn House Office Building. The Subcommittee requires that witnesses deliver 100 copies of their prepared testimony to Room B-349C of the Rayburn House Office Building in advance of the hearing. The copies should be delivered by noon on Tuesday, October 15, 1991.

If you have any questions about the hearing, please contact Robert Gellman of the Subcommittee staff.

Very truly yours,

Bob Wise
Chairman

PRIVACY VIOLATIONS ARISING FROM GENETIC DISCRIMINATION: INFORMING AND LIMITING THE HUMAN GENOME INITIATIVE

Testimony to be delivered by Dr. Paul R. Billings on October 17, 1991 before the Government Information, Justice and Agriculture Subcommittee of the House of Representatives' Committee on Government Operations, Congress of the United States, in Washington, DC.

Rep. Robert Wise of West Virginia, Chairman
Rep. Al McCandless of California, Ranking Minority Member

Mr. Chairman, Congressman McCandless, Members of the Subcommittee, ladies and gentlemen, thank you for the invitation to testify and submit prepared remarks for your consideration concerning privacy and genetic information.

I have appended to this text, which I ask you to include in the Record, several publications and documents which I believe are crucial for your informed review of the topic of this hearing. They include: a paper by Professor Jon Beckwith of Harvard University--the individual who first cloned a human-like gene, a member of the National Academy of Science and a participant in The Human Genome Initiative (HGI) Ethical, Legal and Social Issues (ELSI) Advisory Board--pointing out the limits of genetic investigation and exaggerations which currently characterize the rhetoric of HGI; two publications which outline the significant arguments proposed by members of the scientific community against the organization, research program and funding of HGI; my own research paper describing the results of an investigation of incidents of genetic discrimination which is scheduled to be published in the next few months; a copy of legislation which passed the California State Legislature this year which would have amended the state civil rights statute to include genetic characteristics as protected rights, and limited insurer and employer use of genetic data; and, documents related to this topic prepared by the Council for Responsible Genetics, a private public interest organization which for many years has carefully monitored progress in human genetics and commented constructively on genetic information and public policy issues. I would also commend to you the work of another Boston-based public interest group, the Genetic Screening Study Group (formerly a subgrouping of Science for the People), which through the minutes of its regular meetings, participation in and organization of public forums and scholarly publications has made a substantial contribution to the review of the genetic revolution sweeping biology and medicine.

May I also say before beginning that I received my AB degree in American History from the University of California at San Diego, my MD and PhD (in the field of Immunology) degrees from Harvard University, and my postgraduate education in Internal Medicine and Medical Genetics from the University of Washington. As a faculty member at Harvard Medical School, I conducted basic and clinical research in genetics and immunology and have for the past year been Vice Chairman of the Department of Medicine and Chief of the Division of Genetic Medicine at California Pacific Medical Center, the largest private community-type hospital in Northern California. I am on the clinical faculty of the University of California at San Francisco and a Visiting Scientist at the Human Genome Center (HGC) of Lawrence Berkeley National Laboratories (LBL). I have received financial support from funds appropriated to the HGC at LBL.

I will attempt to keep my comments brief, drawing upon my background outlined above, my work on genetic discrimination and

with individuals presenting with clinical genetic problems or questions. It is sobering and intimidating to note that also testifying today are the co-discoverer of the structure and biochemistry of DNA which encodes human genetic information, a man whose research is directly responsible for the current explosion in genetic information and methods; the co-discoverer of the site of the gene associated with Huntington's Disease (Woody Guthrie's Disease)--an important hereditary adult neuro-psychiatric disorder--who is also the ELSI Advisory Board Chairperson; and important representatives of both the National Institutes of Health (NIH) and the Department of Energy (DOE) who are sponsoring the basic research of HGI and the ELSI Program. It is an exceptionally distinguished and able group you have gathered to provide you with a view of how genetic information and HGI impacts privacy issues; my comments I hope will be found to be useful and distinct.

I will stipulate that genetic research will provide insights into traits and disorders where no reliable basic knowledge currently exists, and therefore is a most hopeful basic scientific endeavor. I will discuss specifically three questions which thus arise:

1. Is the Genetic Age (and HGI) unique?
2. What can we learn about privacy in this country from incidents of genetic discrimination?
3. Will the ELSI Program be effective in preventing violations of individual's right to privacy and the misuse or misapplication of new basic human genetic knowledge?

IS THIS GENETIC AGE UNIQUE?

Rapid advances in our ability to study human traits with genetic approaches and therefore accumulate genetic information about human biology and disease have fostered descriptions of this epoch as the Genetic Age. It is true that finding a gene has become the gold standard of biomedical investigation and publication, that most of the laboratories in- and outside of the NIH are using genetic techniques and that the supporters and officials of the HGI have publically stated that "what it is to be human" and certainly most human disorders, characteristics and behaviors will be explained as a byproduct of the completed HGI.

The absence of clinically relevant experimental systems to study human genetic information, the boring repetitiveness of many molecular genetic observations, their limitations and not infrequent insignificance, and the fact that human genetic insights do not necessarily lead to useful new therapies or preventive strategies is often overlooked or not stated. In addition, it should be clear that for now and the foreseeable future, the major benefit derived from genetic information by families and individuals is the possibility to prevent the birth of other gene carriers by utilizing selective abortion. If abortion becomes practically unavailable to large segments of the American public, the benefits to be derived from HGI will need to

be reconsidered. But is our preoccupation with genetics new?

No. In the 1920's and 30's, in this country and in Europe, the scientific community touted genetic methods and approaches in a similar manner. Genetic data on a wide variety of human traits was published, college and university courses were taught and Congress was lobbied by human geneticists. Genetic explanations for disease and human variation were routinely offered and considered. The rhetoric used during this period--the first Genetic Age--reduced complex individual and societal characteristics to the functional result of gene action. It is strikingly similar to that offered today. This previous Genetic Age waned because the explanatory power of genetic approaches and information was highly limited and primitive, and because citizens including scientists were horrified at the application of genetic information to exclude immigrants, to justify sterilizations of the mentally retarded, psychiatrically ill and alcoholic, to prohibit marriage between racially and genetically distinct groups and to exterminate minorities in other countries. Though the underlying forces which made genetic explanations seductive remained, the science failed and this country's tradition of protecting the individual and civil liberty was asserted. **The first Genetic Age was thus appropriately limited by weaknesses of its science, and by the society it sought to alter.**

Again, we are faced with a period when genetic explanations for individual and social ills are dominant, the misuse of this information to deprive citizens of civil rights and entitlements is occurring, and genetic insights generated in this country are being misused by governments abroad. The science of human genetics (and its methods) is far more sophisticated and successful than that offered during the first Genetic Age--descriptions and predictions about simple genetic traits can be quite accurate and useful. HGI arises well after biomedical science has begun utilizing modern genetic methods to address human traits. It is a result of and gains justification from the progress in studying the genes associated with simple human genetic phenomena, and the wish for explanations of more complex human traits and disorders. It has yet to prove itself useful in providing satisfaction for these inherently human hopes.

In fact, the science of human genetics (and HGI) will succeed in this Age unless it dwells on such traits as the common disorders of modern society, behavioral/mental variation, intelligence, productivity, job performance, creativity, criminality or homelessness. In addition, current conditions for the study of human genetics can be distinguished from the earlier period by the large sums of federal and private investment in the field, the "university/industrial complex" (attributed to Professor Victor McKusick) and the pressure for profitability in the fledgling biotechnology sector. These are new ingredients which may distort or obfuscate the explanatory successes and failures of human genetics and DNA information. **It will likely take a much more comprehensive understanding of science and**

genetics by our society, and a clear nationwide restatement of the value of the individual, and basic social entitlements and civil liberties, for the public to derive the maximal public good from the second Genetic Age.

Thus, the Genetic Age is not unique; the causes of fascination with genetic explanations, the limitations of genetic insights and the priorities our nation must restate to derive the appropriate benefits from the experimental work in human genetics are not new and have not changed.

WHAT CAN WE LEARN FROM STUDYING GENETIC DISCRIMINATION?

The results and origins of genetic discrimination are similar to that justified by racial, gender, ethnic, religious or physical prejudice. In cases of genetic discrimination, real or presumed differences in an individual's genes form the justification for the discriminatory actions and stigma. Since the study of humans by genetic methods highlights human differences (and thus genetics can be said to be a discriminatory science), and some human genetic characteristics and unique genes are common in groups which have historically suffered discrimination in our society, it is not surprising that discrimination as a consequence of genetic information exists. In fact, there are four independent lines of evidence which make clear that genetic discrimination is a current problem in our society:

1. Our society has a penchant for discrimination and prejudice against individuals and groups which arises from perceptions of the broad range of human differences. Human genetic differences, often discovered by federal research programs or identified by state sponsored screening initiatives, offer simply another rationalization for discrimination in our society.

2. When large genetic screening programs have been tried in this country, results have included "redlining", stigmatization, physical harm and confusion. These screening program failures suggest that discrimination exists, and arose when genetic information and understanding were incomplete, when genetic approaches were tried in the face of ambient social inequities and for other yet to be identified reasons.

3. In current practices involving the handling genetic information, there is clear evidence of individuals altering behaviors (adapting) to a discriminatory environment. My research indicates that individuals are "forgetting" to tell physicians, counsellors, insurance agents and employers, hereditary information out of fear of the consequences. Doctors are not making genetic diagnoses to protect the health insurance availability of their patient and the patient's dependents. Insurance agents are advising potential clients to be dishonest on application forms. Individuals who might benefit from genetic testing are declining to be tested in order to preserve their

entitlements--they realize that test information is readily disseminated to employers, insurers, the Armed Forces, other governmental agencies and social institutions. Interestingly, the attestations of normalcy by experts like physicians has little influence on genetic discrimination once begun, which may partially explain the "hiding" adaptation.

4. **Individual cases of genetic discrimination have been reported and verified.** Though it is unclear how common frank discrimination based on the hereditary nature of a trait is occurring, the cases were not difficult to discover. A simple advertisement and interest has generated over 50 cases in two and one-half years of collecting. Most reported incidents involve loss or denial of access to insurances, though discrimination in hiring practices, in availability of adoption and by other social agencies was noted. It was be predicted that genetic discrimination would mark whole families as stigmatized and different since genes "run in families". The collected cases frequently demonstrate the loss of entitlements not only of individuals but spouses and dependents. Illustrative reports include:

--a woman during a routine gynecological check questioned her physician about the possibility of Huntington's Disease in her mother. She subsequently lost all insurances when she applied for life insurance and her medical records were reviewed.

--an individual with a mild, stable, barely perceptible genetic characteristic applied for life insurance. His doctor attested to his good condition while sending his medical records to an insurance broker, who serviced all this individual's insurance needs. Not only did he not get his life insurance, but his auto insurance was canceled; a real problem for this travelling salesman without a traffic ticket or accident in 20 years of driving.

--a police department heard a rumor of a genetic disorder in a academy graduate and queried him. When he admitted this family history, he was told to undergo genetic testing or not be hired.

--whole families are excluded from insurances when one boy has a genetic form of mental retardation, even when this does not affect other aspects of his health and has no significant manifestations in females. Whole families are marked by a single test.

--information concerning a family history of a hereditary disorder, either developed by private investigation or confession, has been used as exclusion criteria for adoption services.

--individuals and families who were unaware of giving "informed consent" for their inclusion in state sponsored genetic screening

programs have nonetheless been identified by these initiatives and excluded from insurances because of "pre-existing" conditions.

--individuals who require expensive treatments may be challenged to prove they did not know they had these genetic conditions in the past. They are labelled by requesting treatment.

--individuals with mild, remitted or treated genetic traits or conditions may have record in the Medical Information Bureau, Inc., are usually unaware of its existence or correctness, and thus may not be able to get any insurances.

Several other observations should be mentioned. Self insuring employers or those who collaborate with insurance companies to administer benefits can come in to possession of personal genetic information about employees and may store this data at the workplace. Because physicians must include diagnostic information in order to submit bills for payment to third parties (and state or federal agencies), this appears to be the most common route of privacy violations of genetic information. Finally, given these billing practices and the ubiquitous reviews of medical charts by staff in physician offices, HMOs, insurance agencies, workplaces, and at other sites, the privacy and confidentiality of genetic information found in medical charts is neither apparent nor real.

To summarize, genetic discrimination exists, is frequently associated with violations of the privacy of personal genetic information and results in the loss of the entitlement of access to adequate health care, financial safety, employment opportunity and other social benefits.

WILL THE ELSI PROGRAM BE EFFECTIVE IN PREVENTING PROBLEMS?

The ELSI Program has distinguished itself in several ways. First, it is likely the largest program in biomedical ethics ever, much as HGI is the largest single human biological research program now receiving federal funding. Yet as HGI arises from a turn to genetics in biomedical research over the last two decades, so too the ELSI Program follows from the growing call for ethical and social evaluations of science's impact on medicine and society. The appropriate relationship between social and ethical evaluation and basic scientific initiatives and applications is not obvious. Though the history of genetics suggests problems with the interpretation and application of basic data in our society (issues which might be amenable to ELSI type research), how and when proper ethical restraints will be identified, and their influence on the basic HGI program and its applications is uncharted territory. A social mandate on civil rights may be easier to arrive to generate than a clear conclusion from ELSI directed research programs. Nonetheless, the commitment of approximately 100 million dollars over the

course of HGI should stimulate research and insights into the problems of applying genetics to our society. The ELSI Program, with its important issues and difficult research agenda, probably requires a significantly higher fraction of the HGI budget than the three percent originally offered.

Second, the ELSI Program was formulated and initiated at about the same time as the basic science program of HGI was organized. This gives the impression that ethics might temper or modify science not just follow and explain its results and costs. As yet, there is little evidence of this influence.

Third, the Program has recruited an excellent Advisory Panel (of consultants), administrative staff and actively sought to review research relevant to HGI and its applications. Yet to be effective it will have to marshall social, economic and political forces in order to influence the course of genetic inquiry and improve the outcomes of any applications attempted. It is unclear whether the current program is constituted, empowered and independent enough to generate the political influence and policies which may be required.

It <u>is</u> quite clear that the ethical, social and legal issues which the application of genetic information in our society highlights are generally not unique. Problems with our entitlement systems, in protecting civil rights and in the conduct of science and federal programs occurred well before the ELSI Program. For instance, issues which existed before the ELSI Program but which are relevant to its success include:

1. Universal access of adequate health care and financial safety irrespective of any test results or pre-existing traits.

2. The right of the individual to maintain his or her identity and personal health information as confidential.

3. The right of the individual to have autonomy and choice in health matters even when this may oppose public health needs.

4. The influence of public and social policy on scientific freedom.

5. The proper role and personal limitations of basic scientists as "experts" and "advisors" on the application of their basic data in clinical and social settings.

6. The pervasive problem of scientists who have a conflict of interest in the assessment of the appropriate application of their basic data. (For instance, the discoverer of a gene may be more likely to claim that a clinical test should be developed if the scientist either owns or receives funding from a genetic testing private enterprise).

As indicated, these problems and those which produce genetic discrimination are not caused by HGI and the explosion of genetic information but may need to be fixed before human genetic testing

and information becomes more prevalent in our society. If this is recognized as a necessity, it is unclear that an organization of the sort that the ELSI Program represents will be adequate or effective in generating these changes.

In conclusion, I will suggest certain explicit modifications in the current ELSI Program, to include:

1. **The ELSI Program should be constituted as a separate activity from the basic research program of the HGI (and receive a higher proportion of its allocation)**, and should be organized in such a manner that if the occasion arises, it can act freely and independently to monitor and restrict the application or conduct of HGI research and effectively generate public policy initiatives to improve HGI related outcomes and achieve limitations if necessary.

2. Given the personal nature of genetic information, and its inherent relationship to reproduction and other family members, **the ELSI Program should consider immediate initiatives to not only limit the discriminatory uses of genetic information but prohibit its collection, storage and evaluation by any individual or social agency aside from the specific individual from which it is derived. The elimination of access to such information is the only enforceable means to insure its privacy.**

3. The ELSI Program should adopt a statement and policy program recognizing that **certain social conditions may be worsened in this country by the widespread application of genetic information now and thus, this may preclude applications and delay the possible benefits which might be derived by the public from HGI. Clear statements about the rights, entitlements and importance of funding of research into individuals already born with genetic disabilities is required. Similarly, it should be stated that social, political and cultural differences in other societies may make the free exchange of genetic information and methods developed by HGI with these societies unadvisable or harmful.** The ELSI Program in concert with HGI and individual scientists must make these determinations in evaluating which of our research programs to conduct and the type of international collaborations to allow.

4. **The ELSI Program should insist that all individuals participating in HGI activities and funded research make full and public disclosures of private contracts or holdings in companies utilizing the results of genetic research or genetic information.**

5. **A statement and policy indicating that HGI research which may harm individuals, can not be done in a manner which is verifiable and yields useful basic information or which is an improper use of limited research funds could be banned by ELSI Program action. At present, this might include experiments in human germline gene therapy.**

In conclusion, the genetic revolution and HGI will exacerbate developing problems impinging upon and limiting the individual's right to privacy in our society. Public education and participation is essential to the proper evaluation of HGI and for setting policy priorities as changes and threats to civil liberty traditions arise. Though the ELSI Program may develop research data which will illuminate many issues, it is unclear whether its current organization, mandate and funding will allow it to effectively evaluate, monitor and restrict (if necessary) the conduct of basic aspects of HGI. Furthermore, a broad social referendum restating the basic rights of Americans to civil rights, privacy and social entitlements, and adequate social programs and enforced laws to insure this environment may be needed before the results of HGI can be enjoyed by all citizens.

Remarks of Philip Reilly, M.D., J.D.

On Behalf of the American Society of Human Genetics

to

The Congress of the United States

Testimony before the Government Information,

Justice and Agriculture Subcommittee

of the Committee

on Government Operations

October 17, 1991

Statement on Genetics and Privacy

Good morning. My name is Philip Reilly. I am the Executive Director of the Shriver Center for Mental Retardation in Waltham, Massachusetts. I am grateful for the opportunity to speak to you today about a topic that I have been studying for nearly twenty years: genetic testing and how it impacts on the right to privacy.

I also have the honor to appear before you as a representative of the American Society of Human Genetics (ASHG), the membership of which includes 4500 medical geneticists, genetic counselors and scientists. I chair the Social Issues Committee of the ASHG. No group in this nation has thought more about the problems you are exploring than has the ASHG. The Board of Directors of our Society has authorized me to speak to you on its behalf.

Dramatic advances in molecular biology coupled with an ever growing awareness that much of the burden of human disease (including cancers, heart diseases, diabetes and dementias) has a genetic component set the stage for the Human Genome Project. Although daunting in size, the goal of sequencing the entire human genome will succeed. One immediate and crucially important consequence of the Human Genome Project will be an avalanche of new diagnostic, presymptomatic and predispositional tests for genetic and genetically influenced disorders. Our ability to ascertain those at risk will for some years exceed our ability to treat persons with these

disorders. Nevertheless, there will be profound interest in testing for various genetic conditions in the hope of avoiding or ameliorating them.

Systematic genetic testing in this nation began 30 years ago with the introduction of newborn screening programs to identify children with genetic diseases such as phenylketonuria (PKU), which causes severe mental retardation unless treated with a special diet. Today, nearly all newborns in the US are screened for several genetic disorders that if identified early are preventable or subject to amelioration. For example, over 99 percent of the 550,000 children born each year in California are tested for three genetic conditions. These state based (legislatively mandated) programs collect and store millions of bits of genetic data each year!

Our experience with genetic screening for persons who carry a gene that, depending on whom they marry, may pose a high (1:4) risk of bearing a child with a serious disorder has been imperfect. Screening for Tay-Sachs disease (a fatal neurodegenerative disease of childhood) carrier status has been quite successful. Since 1971 when Tay-Sachs carrier screening began there has been more than a 90% reduction in the annual number of infants newly diagnosed with Tay-Sachs disease. In contrast, programs developed in the early seventies to screen black persons for sickle cell trait caused significant social harm to that group. For a time persons found to carry one copy of the sickle cell gene experienced unjustified discrimination in the workplace and among insurers. It is noteworthy that sickle cell testing was often conducted pursuant to laws that mandated screening. The errors of those laws should not be forgotten. The major event that corrected the unintended injuries caused by sickle cell screening programs was the enactment in 1972 of a

federal law that conditioned the award of funds for screening to states upon proof of adherence to certain fundamental principles: that genetic testing must always be voluntary, that all persons must have access to adequate pre-test education and post-test genetic counseling, and that genetic information must be considered confidential.

Just as our investigation of the human genome is beginning to generate many new diagnostic tests, so public concern about genetic discrimination is stimulating much legislative interest in preventing misuse of data derived from such tests. On behalf of the ASHG, I informally monitor new bills that pertain to this matter. During the last two years there has been much legislative activity. Many states have enacted laws creating forensic DNA data banks, a large number have enacted laws to permit the use of DNA test information to resolve paternity disputes and to aid in the resolution of crimes of violence, and several are debating bills to regulate or prohibit the use of genetic test information by insurers and employers.

Clearly, genetic data derived primarily for clinical purposes are likely to stimulate significant interest among third parties. This raises a number of privacy concerns . I shall divide my comments on genetics and privacy into two sections: those that involve family members and those that involve insurers, employers and other social institutions.

First, unlike classical medical information such as the diagnosis of pneumonia, genetic data derived from a single individual may be of great importance to other family members. For example, if a physician diagnoses Fragile X syndrome, a quite common heritable form of mental retardation,

that fact may be of great potential relevance to the reproductive plans of the patient's aunts and sisters. The principle of doctor-patient confidentiality holds that the physician should not disclose test data to third parties. But, if a patient does not share genetic information, the physician may be placed in the quandary of knowing that there is a relative who is at high risk for a genetic problem, but of not having an ethically acceptable means of alerting that person to this problem. The fact that genetic testing of an individual often yields facts of great importance to other persons may lead us to modify our thinking about the boundaries of doctor-patient confidentiality.

Second, we live in a society where persons with chronic diseases have trouble obtaining work and adequate health insurance and life insurance. Given that we are poised on the threshold of an era when we may vastly expand our understanding of whom is likely to become chronically ill, this is especially troubling.

Currently, neither insurers nor employers make substantial use of genetic test results, but life insurers do routinely use related information. For example, anyone who purchases a moderately large whole life insurance policy is, likely to undergo cholesterol testing. Tests for diabetes mellitus and high blood pressure, both strongly influenced by genetic factors, have long been used as part of the qualification for both life and health insurance. Some of those who are discovered to have elevated cholesterol, urinary sugar, or high blood pressure may have trouble securing the coverage they seek. Similarly, the insurance applicant is often asked to provide information on family history. People who disclose a substantial history of familial

cancer may pay higher premiums, be denied coverage for specific disorders or be denied coverage altogether. Someday soon we may be using genetic tests to assess one's risk of developing cancer. Persons who learn that they are at increased risk are likely to have significant difficulty obtaining life insurance at reasonable rates.

Human genetics which is, in essence, the study of biological variation within our species, teaches that each of us carries genes which may be harmful to our health or in our reproduction. It would be a tragedy indeed if the scientific achievements of the Human Genome Project were eclipsed by institutional practices that disenfranchised thousands of Americans from the workplace or denied them affordable insurance to safeguard their families.

The American Society of Human Genetics applauds the growing Congressional and state legislative interest in the privacy problems that surround genetic data. The Society has reviewed HR 2045, the proposed Human Genome Privacy Act, and regards it as an important initial effort that with further study and revision should provide an important basis for the protection of privacy.

The ASHG believes that any plan to guard the privacy interests of persons undergoing genetic tests must:

(1) Proceed on the premise that unauthorized disclosure of genetic data to third parties may seriously harm the individual who has been tested.
(2) Determine who should be authorized to collect genetic information,

how it should be stored, how it may be linked to other data, who should control access to it, who should have access to it, and how such data may be used or released.

(3) Develop rules that clearly define the permissible and impermissible uses of such data by third parties such as insurers, employers, and school systems.

(4) Place the burden on those who would use genetic data to limit access to insurance, employment or other social institutions to provide scientifically rigorous justification for that decision.

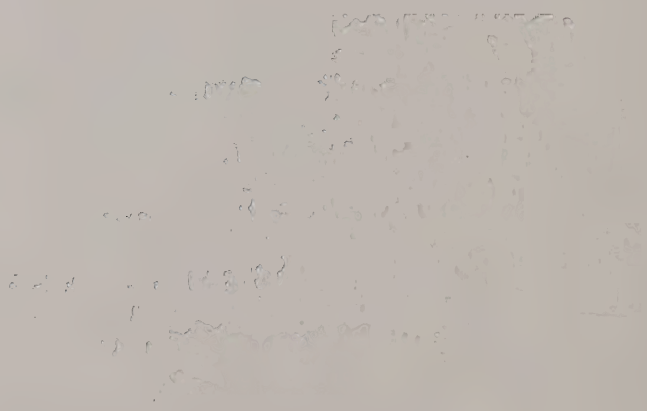
(5) Recognize that it is important to permit qualified researchers with legitimate protocols to gain access to genetic data banks so long as the information therein is studied anonymously. (This is essentially the view taken by The Privacy Protection Commission in its 1977 Report.)

(6) Characterize the violation of genetic data banks and wrongful collection, use or dissemination of genetic data as a criminal act and, also, create civil remedies for persons harmed by wrongful disclosure.

(7) Understand that the highest priority should be given to developing innovative efforts to educate our citizens about genetics.

Advances in understanding the human genome promise immense benefits to humanity. As is the case with all new technologies, it is our collective task to introduce genetic tests (and, in time, genetic therapies) in a manner that causes the least possible harm. The best way to achieve this is to greatly increase educational efforts about human genetics in the United States. The dividends from so doing might be rich indeed. It takes but a moment's reflection to realize that racism, ethnic strife and discrimination on the basis

of gender, among our most intractable and debilitating problems, are in part due to misperceptions about relatively minor genetic differences. A society whose members understood that genetic diversity is completely compatible with the principle of human equality would have little incentive to misuse genetic data. Thank you.



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STATEMENT
OF
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PRESIDENT OF THE FOUNDATION ON ECONOMIC TRENDS
WASHINGTON, D.C.
BEFORE THE SUBCOMMITTEE ON GOVERNMENT INFORMATION,
JUSTICE AND AGRICULTURE
UNITED STATES HOUSE OF REPRESENTATIVES

October 17, 1991

Mr. Chairman and members of the Subcommittee. In the 1950s and 60s we saw the growth of the civil rights movement in the U.S. In the 1970s and 80s a national and international human rights movement developed that profoundly altered Eastern Europe and ended the Cold War. In the 1990s a powerful new genetics rights movement will emerge in countries around the world -- a movement which will force governments to address the issues of genetic discrimination and eugenics raised by advances in biotechnology.

Genetic screening is a powerful new diagnostic tool with the potential for great benefits and great harm. Humankind is afflicted by more than 3,000 inherited diseases. Today, we have identified fewer than 3 percent of all known inherited disorders. There is little doubt that maps of the human genome would greatly facilitate the search for genes related to specific inherited diseases. Additionally, the ability to determine whether individuals are carriers of specific gene defects will facilitate investigations of the disease risks associated with certain occupations, drugs and environmental problems.

New genetic screening technologies could also have a profound negative impact on many constituencies, including workers, women, the disabled, minorities, health care consumers, physicians, insurance consumers and others. These groups are already concerned that genetic screening information could be used as a tool of discrimination and eugenics based on genetic makeup. Individuals with so-called "superior" genetic readouts could achieve social advantage in decisions ranging from conception and education, to job choices and insurance availability.

Genetic information about an individual's pre-disposition to a variety of physical or mental disorders is the most intimate and private data about ourselves. The Dupont decision to screen Afro American workers for sickle-cell anemia, in the 1970s, was greeted with alarm by many in the black community. The sickle cell anemia fiasco also showed how discriminatory and damaging the use of genetic technology can become. Government studies indicate that many Fortune 500 companies are interested in using genetic testing to screen workers for a variety of predispositions in order to minimize liability for injuries and disease incurred in the work place. As we look towards wide-spread screening of employees, we have to ask a fundamental question about our priorities: do we screen or genetically alter workers to fit an unhealthy workplace, or do we change the workplace into a healthier human environment.

There has also been increased concern about how insurance companies will utilize the new genetic screening technology. Insurance companies could establish policies requiring those with certain genetic predispositions to pay higher rates. For example, an auto insurance company could force applicants to be tested for a pre-disposition to alcoholism and refuse coverage for such individuals or force them to pay higher premiums.

Such a policy would discriminate against those with the "alcohol" gene even though they may never become alcoholics.

Others are concerned that our educational system will begin tracking children based on their genetic predispositions for intelligence or learning aptitude. Those who find such a scenario far fetched should be aware that last month the National Institute on Child Health and Human Development (NICHHD) announced that it was sponsoring a $600,000 program to explore the genetic links to I.Q. The possible discriminatory use of such programs is obvious.

Government agencies are also getting involved in genetic screening and identification. The National Institutes of Health (NIH) has announced that it is planning pilot programs to test the feasibility of nation wide genetic screening for the cystic fibrosis gene. Many scientists have already voiced strong objections to this project viewing it as "incredibly premature" and potentially discriminatory. The Department of Defense has recently announced that it now intends to maintain DNA "dogtags" on all armed forces personnel. As millions of Americans become subject to government mandated genetic identification and screening, genetic privacy will become a national priority.

Equally disturbing is the growing trend in pre-natal diagnosis. Using amniocentesis and chorionic villus testing it is now possible to gain a genetic readout of fetuses prior to birth. Parents are totally unprepared for the flood of new information on their offspring that will flow from the work of the human genome project. A couple's decision to abort could become totally dependent on a genetic counselor's description of the importance or magnitude of a genetic predisposition. A recent poll showed that 11% of Americans might abort a fetus pre-disposed to obesity. Pre-natal screening could lead to a new and virulent

form of eugenics -- one based not on political philosophy or the whims of a dictator but rather driven by the fears of parents and the potential for a multi-billion dollar screening industry.

The extraordinary discriminatory and eugenic potential of genetic diagnosis and screening has been debated over the last two decades. In 1977, I participated in a demonstration at the National Academy of Sciences to force that institution to consider the social and ethical implications of genetic engineering. Over the last fourteen years we have appeared at numerous NIH Recombinant DNA Advisory Committee meetings attempting to convince that agency to take a more serious look at the moral implications of their work. When necessary we have petitioned and even litigated to ensure responsible regulation of genetic screening and human gene therapy. Throughout, the NIH and the other agencies have been recalcitrant and even hostile to considering the non-scientific aspect of advances in biotechnology.

As early as 1978, I met with thirty members of the House of Representatives at a Congressional Clearinghouse for the Future Meeting to discuss the implication of genetic engineering. I remember Congressman Peter Rodino suggesting the need for a comprehensive legislative strategy to deal with genetic screening and related issues. Now, it is thirteen years later, and still not a single piece of legislation regulating genetic engineering has passed the Congress.

We are hopeful that things may be changing. We are extremely grateful to Representative Conyers for introducing historic genetic privacy legislation. The Human Genome Privacy Act extends the right of privacy to include genetic information. It applies the civil rights standards legislated and fought for in the 60s and 70s to the genetic code.

Congressman Conyer's bill requires the federal agencies and their contractors and grantees to assume leadership in the crucial area of genetic privacy. It is the first piece of a series of legislative initiatives that will be required to ensure that genetic information is used for our benefit and not as a new and pernicious form of discrimination.

We welcome the generally favorable reception that this legislation has received in the scientific community and at the federal agencies. Individuals in the molecular biology field, such as Dr. French Anderson, have publicly stated their support for genetic privacy legislation and have urged government regulation against the discriminatory and eugenic use of their work. Unfortunately, many in the biotechnology field have remained silent on the need for legislative oversight of their field. Cooperation between the emerging genetic rights movement and the scientific community is essential if we are to effectively prevent abuses of genetic screening in the workplace, in health and insurance coverage and throughout the public and private sector.

Unlike many other activist groups around the world, our organization has not opposed appropriations for the Human Genome Project. Despite national and international pressure from many of the groups we work with, we have continued to support the growth of the human genome initiative. However, this support is given with one major qualification -- continued appropriations for the genome project must be accompanied by promulgation of the appropriate legislative safeguards against the abuse and misuse of genetic screening. Should the Congress fail in establishing legislative safeguards, we will actively seek a moratorium on all research activities of the Human Genome Project.

It will take the combined energies of a great number of Americans to attain passage of the Human Genome Privacy Act and subsequent legislative efforts required to deal with

the social challenges presented by advances in the genome project. In order to garner this crucial political support, we are announcing today that our organization will be hosting a conference, in early 1992, bringing together leaders from a wide range of constituencies from fifteen countries to launch a genetics rights movement in the U.S. and Europe. The principle focus of this first international "Genetic Rights" convention will be to address the critical discrimination and eugenic issues surrounding advances in the Human Genome Project.

We are also announcing today the formation of a national legal network to represent workers, women, the disabled and minorities and others who are victims of genetic discrimination. This network will be similar to those set up in the 1950s and 60s for civil rights and will provider pro bono legal help for those suffering genetic discrimination at the hands of private corporations or the government.

Thank you, Mr. Chairman, for this opportunity to testify.